



First Master Class in Skeletal dysplasias

MARCH 9-10-11, 2026,

Imagine Institute, Paris

This course aims at delivering up-to-date knowledge on skeletal dysplasias incorporating emerging technologies to: clinical and molecular geneticists in training or certified, genetic counsellors, pediatricians, radiologists, rheumatologists and orthopedic surgeons. It creates a real opportunity for interaction and discussion with experts from all over Europe and learn how to work in a network environment in this challenging and growing field of expertise. The faculty combines experts from radiology and clinical and molecular genetics from 5 different countries.

The presentation of “real life” cases presented by the faculty staff, from the observation of a single major skeletal feature and then discussing how to arrive to the diagnosis, will favor interactive discussion and practical teaching. Participants are also encouraged to present a skeletal dysplasia case (solved or unsolved) for on-site discussion and will learn how to use the CPMS 2.0 platform for ERN BOND online case discussion.

PROGRAM

Monday, March 9, 2026 (Day 1)

13h00-13h30	Registration & Welcome coffee
13h30-14h00	Introduction : the field of skeletal dysplasia in 2026 Valérie Cormier-Daire, Paris
14h00-15h	Place of the clinical approach Geert Mortier, Leuven
15h00-16h00	Radiological approach and the digital era Alistair Calder, London
<i>Coffee Break</i>	
16h30-17h30	Incorporating innovative technologies in the diagnostic pathway of skeletal dysplasia

Karen Heath, Madrid

17h30-18h30 Challenges the prenatal diagnosis of skeletal dysplasia

Valérie Cormier-Daire, Paris

Free time for evening meal in Paris

Tuesday, March 10, 2026 (Day 2)

08h45-09h45 Principles of management and novel perspectives in treatment.

Sergio Sousa, Coimbra

09h45-10h45 **Keynote Speaker : Segmental overgrowth.**

Guillaume Canaud, Necker enfants malades hospital, Paris

10h45-11h15 Coffee Break

11h15-12h30 Case Presentations by **faculty staff** : Prenatal and neonatal

1) Bowing of the femur

2) Short femur

12h30-13h30 LUNCH

13h30-15h30 Case Presentations by **faculty staff** :

1) Child with short trunk (disproportionate short stature)

2) Bone fragility and fractures in children

15h30-16h00 Coffee Break

16h00-18h00 Case Presentations by **the attendees**. 8 presentations (solved cases)

18h00-19h00 Quiz (pathonomic signs and differential diagnosis, with refreshment.

Official course dinner for all

March 11, 2026 (Day 3)

08h45-10h00 Case presentations by **the attendees**. 5 presentations (unsolved cases)
CPMS 2.0 hands on demonstration

10h00-10h30 Coffee Break

10h30-12h30 Case presentations by **faculty staff** – complex cases :

1) Phenocopies

2) Reverse phenotyping (VUS and what to do)

3) Negative genetic NGS – what to do?

12h30-13h30

LUNCH

Departure

Number of participants : 25

Fees (include welcome coffee, 4 coffee breaks, 2 lunches, refreshment during the quiz and the official dinner) :

100 euros for Trainees / 180 euros Regular

ESHG fellowships (EUR 600) are available for ESHG members.

If you wish to apply for a fellowship, please write half a page documenting your name, job title, location, and justification to valerie.cormier-daire@inserm.fr. Please note that there are 10 fellowship tickets available.

Contact data of applicant to be sent at Valerie Cormier-Daire

- Name

- Address

- Email

- Phone

-CV/ Letter of motivation

Contact : Cormier-Daire, Valerie, Necker enfants malades Hospital and Imagine institute

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