

2025 ESPE-OSCAR SCIENCE SYMPOSIUM

Mineralization of bone and growth plate,
towards the development of new therapies

PARIS
September
18 & 19



PROGRAMME

ESPE
European Society for
Paediatric Endocrinology

OSCAR
FILIÈRE
SANTÉ
MALADIES
RARES
filieres-oscar.fr

SCIENTIFIC AND LOCAL ORGANIZING COMMITTEE

ESPE-OSCAR
PARIS 2025



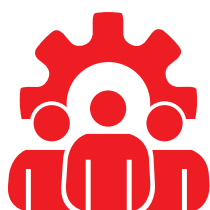
Justine BACCHETTA (Lyon, France)
Claire BARDET (Paris, France)
Karelle BENISTAN (Garches, France)
Karine BRIOT (Paris, France)
Mireille CASTANET (Rouen, France)
Roland CHAPURLAT (Lyon, France)
Catherine CHAUSSAIN (Paris, France)
Martine COHEN-SOLAL (Paris, France)
Valérie CORMIER-DAIRE (Paris, France)
Maud DE DIEULEVEULT (Paris, France)

Thomas EDOUARD (Toulouse, France)
Isabelle GENNERO (Toulouse, France)
Pascal HOUILLIER (Paris, France)
Hervé KEMPF (Nancy, France)
Gilles LAVERNY (Strasbourg, France)
Sandrine LEMOINE (Lyon, France)
Agnès LINGLART (Le Kremlin-Bicêtre, France)
Benoit MIOTTO (Paris, France)
Julien VAN GILS (Bordeaux, France)
Frédéric VELARD (Reims, France)

The European Society for Paediatric Endocrinology (ESPE) is an international non-profit scientific organisation dedicated to improving the clinical care of children and adolescents with endocrine and metabolic conditions, including diabetes and rare bone disorders, through research, education, and the development of clinical standards. ESPE is a founding member of the International Consortium of Pediatric Endocrinology (ICPE) and actively promotes global collaboration in the field. Its mission is to advance excellence in paediatric endocrinology, diabetes, and bone disorders by fostering scientific discovery, medical education, and high-quality clinical care.

OSCAR - French rare diseases Healthcare Network: bone, cartilage and calcium diseases, officially designated and funded by the French Ministry of Health since 2014 under the National Rare Disease Plan. OSCAR is hosted by Assistance Publique – Hôpitaux de Paris (AP-HP) and coordinates a national network of expert centers, patient associations, research laboratories, and healthcare professionals dedicated to improving care, research, and education in rare skeletal diseases.

This event has been submitted for accreditation to the European Accreditation Council for Continuing Medical Education (EACCME®). Final credit assignment depends on verified attendance.



ORGANIZER

European Society of Pediatric Endocrinology (ESPE) & OSCAR, The French network of rare bone, calcium and cartilage diseases

EVENT

**2025
ESPE-OSCAR
SCIENCE SYMPOSIUM**

DATES

September 18 & 19, 2025

VENUE / CITY

**PARIS – FRANCE
Hyatt Regency Paris Étoile**

SPEAKERS & CHAIRS



A

Jakub ABRAMSON

PhD, Associate Professor Head of Abramson Lab "Immune tolerance & Autoimmunity", Faculty of Biology, Department of Immunology and Regenerative Biology, Weizmann Institute of Science, Rehovot, Israel

Inês ALVES

DVM, PhD (candidate) – Founder & President, ANDO Portugal; Patient Representative, COMP-EMA & ERN BOND; Co-leader, EuRR-Bone Outcomes; EUPATI Portugal Vice-President; PhD Candidate in Human Kinetics, University of Évora & CHRC, Évora, Portugal

Thomas Levin Geiser ANDERSEN

PhD, Associate Professor, Department of Clinical Research, University of Southern Denmark, the Department of Pathology, Odense University Hospital, and the Department of Forensic Medicine, Aarhus University in Denmark. Head of Molecular Bone Histology Lab (MBH Lab), and the Danish Spatial Imaging Consortium (DanSIC), Odense, Denmark

Natasha APPELMAN-DIJKSTRA

MD, PhD, Professor of Internal Medicine section Endocrinology, Head of Center for Bone Quality, Leiden University Medical Center, Chief Educator of Internal Medicine for the Leiden education region, Leiden, The Netherlands

Salome BATSASHVILI

Endocrinology Fellow, LLC Aversi Clinic, and lectures at European University, Tbilisi, Georgia

B

Justine BACCHETTA

MD, PhD, Professor of Pediatric Nephrology, Paediatric Nephrology Rheumatology and Dermatology Unit, Reference Center for Rare Diseases of Calcium and Phosphate Metabolism, Hospices Civils de Lyon, Claude Bernard Lyon 1 University, INSERM UMR1033 Research Unit, Lyon, France

Claire BARDET

PhD, HDR, Professor, Head of Orofacial Rare Diseases Research Group, INSERM UMR 1333 Oral Health, Vice-Dean Research UFR Odontologie, Faculty of Health, Paris Cité University, Paris, France

Martin BLOSSE DUPLAN

DDS, PhD, Professor of Periodontology, Oral Medicine Department, Reference Center for Rare Diseases of Calcium and Phosphate Metabolism, Bretonneau Hospital AP-HP, Paris Cité University, Lab Oral Health, INSERM UMR1333, Paris, France

C

Roland CHAPURLAT

MD, PhD, Professor of Rheumatology, Head of Rheumatology and Bone Diseases Department, Reference Center for Fibrous Dysplasia of Bone, Hospices Civils de Lyon, Claude Bernard Lyon 1 University, Head of INSERM UMR1033 LYOS Research Unit, Lyon, France

Catherine CHAUSSAIN

DDS, PhD, Professor of Odontology, Oral Medicine Department, Reference Center for Rare Diseases of Calcium and Phosphate Metabolism, Bretonneau Hospital AP-HP, Paris Cité University, Lab Oral Health, INSERM UMR1333, Paris, France

Martine COHEN SOLAL

MD, PhD, Professor of Rheumatology, Rheumatology Department, Reference Center for Rare Bone Diseases, Lariboisière Hospital AP-HP, Paris Cité University, Head of INSERM UMR 1132 BIOSCAR Research Unit, Paris, France

Valérie CORMIER-DAIRE

MD, PhD, Professor of Medical Genetics, Genomic Medicine for Rare Diseases Department, Reference Center for Rare Bone Diseases, Necker Enfants-Malades Hospital AP-HP, Paris Cité University, INSERM UMR 1163, Imagine institute, Paris, France

E

Thomas EDOUARD

MD, PhD, Professor of Pediatrics Endocrinology, Head of Paediatric Endocrinology and Bone diseases Unit, Reference Center for Rare Diseases of Calcium and Phosphate Metabolism, Filière OSCAR, ERN BOND, Children's Hospital of Toulouse, University of Toulouse, Toulouse, France



SPEAKERS & CHAIRS

**Alexandra ERTL**

MD, Pediatric Endocrinologist, Reference Center for Rare Diseases of Calcium and Phosphate Metabolism, Bicêtre Hospital AP-HP, Le Kremlin-Bicêtre, France

F**Brian FOSTER**

MS, PhD, Associate Professor, Division of Biosciences, College of Dentistry, The Ohio State University, Ohio, USA

Nadja FRATZL-ZELMAN

PhD, HDR, Senior Researcher, Ludwig Boltzmann Institute of Osteology at the Hanusch Hospital of OEGK and AUA Trauma Centre Meidling, 1st Medical Department, Hanusch Hospital, Vienna Bone and Growth Center, Vienna, Austria

G**Frédéric GAULOIS**

President of the association of patients and families affected by Hypophosphatemic Vitamin-Resistant Rickets or X Linked Hypophosphatemia (RVRH-XLH), Suresnes, France

Corinna GRASEMANN

MD, Professor of Pediatrics Endocrinology, Head of the Center for Rare Diseases (CeSER) at the University Children's Hospital in Bochum, Ruhr University Bochum, Bochum, Germany

H**Dieter HAFFNER**

MD, Professor of Pediatrics, Head of Department of Pediatric Kidney, Liver and Metabolic Diseases, Hannover Medical School (MHH), Director, KfH Kidney Center for Children and Adolescents, Hannover, Germany

Karen E. HEATH

PhD, ErCLG, Director of the Institute of Medical & Molecular Genetics (INGEMM), Head of the Skeletal dysplasia laboratory, INGEMM, Hospital Universitario la Paz, IdiPAZ; Skeletal dysplasia multidisciplinary Unit (UMDE, ERN BOND), and CIBERER, ISCIII, Madrid, Spain

Wolfgang HÖGLER

MD, FRCPCH, Professor of Paediatrics, Chief of Paediatrics, Centre of Growth and Osteology, Department of Paediatrics and Adolescent Medicine, Johannes Kepler University Linz, Linz, Austria

K**Hervé KEMPF**

PhD, INSERM Researcher, Head of the Research Team CARPATH (Cartilage Pathophysiology and Therapeutics) at the UMR 7365 IMoPA (Molecular & Cellular Engineering and Pathophysiology) Laboratory, CNRS, University of Lorraine, Nancy, France

Uwe KORNAK

MD, PhD, Professor University of Göttingen, Göttingen, Germany

L**Anne-Laure Leyla LAKHEL**

DMD, PhD candidate – Interregional Department of Oral Medicine, AP-HP (Assistance Publique-Hôpitaux de Paris); Paris Cité University, Lab Oral Health, INSERM UMR1333, Paris, France

Gilles LAVERNY

PhD, HDR, Researcher INSERM, Head of a Research Subgroup on the Pathophysiological role of vitamin D signaling, at the Institute of Genetics and Molecular and Cellular Biology, IGBMC, CNRS UMR 7104/INSERM U1258, Strasbourg University, Strasbourg, France

Laurence LEGEAI-MALLET

PhD, HDR, Director of Research INSERM, Head of INSERM UMR 1163 Research Unit, Imagine Institute, Paris, France

Agnès LINGLART

MD, PhD, Professor of Pediatrics, Head of Paediatric Endocrinology and Diabetology Department, Reference Center for Rare Diseases of Calcium and Phosphate Metabolism, and Filière OSCAR, Bicêtre Hospital AP-HP, Paris Saclay University, INSERM, Le Kremlin-Bicêtre, France



SPEAKERS & CHAIRS

**M****Arnaud MOLIN**

MD, PhD, MCU Medical Genetic, Medical Genetics Department, Reference Center for Rare Diseases of Calcium and Phosphate Metabolism, University Hospital of Caen, UR 7450 Biology, genetics and osteoarticular and respiratory therapies (BIOTARGEN) Research Unit, University of Caen Normandy, Caen, France

O**Nicolas OBTEL**

DDS, PhD candidate, Senior Clinical Assistant, Charles-Foix Hospital AP-HP, Paris Cité University, Lab Oral Health, INSERM UMR1333, Paris, France

P**Raja N. R. PADIDELA**

MD, Professor, Consultant Paediatric Endocrinologist & Metabolic Bone Diseases, Head of the Children's Bone and Mineral Disorder service, Royal Manchester Children's Hospital, The University of Manchester, Manchester, United Kingdom

Reena PERCHARD

MBChB MRCPCH PhD, NIHR Academic Clinical Lecturer, Division of Developmental Biology & Medicine, Faculty of Biology, Medicine & Health, University of Manchester, Royal Manchester Children's Hospital, Manchester, United Kingdom

Aikaterini PEROGIANNAKI

MD, Department of Paediatric Endocrinology, University College Hospital, London, United Kingdom

R**Adalbert RAIMANN**

MD, Pediatrics and Adolescent Medicine, Comprehensive Center for Pediatrics, Department of Pediatrics and Adolescent Medicine, Medical University of Vienna, Vienna, Austria

Mara RIMINUCCI

MD, PhD, Professor of Anatomic Pathology, Department of Molecular Medicine Sapienza University of Rome, Rome, Italy

Virginia ROSSI

MD, Paediatric Resident, Department of Paediatric Endocrinology, Royal London Hospital, Barts Health NHS Trust, London, United Kingdom

Frank RUTSCH

MD, Professor of Pediatrics, Head of the Metabolic Department, Muenster University Children's Hospital and Center for Rare Diseases, Muenster University Hospital, Muenster, Germany

V**Johan SERGHERAERT**

MCU-PH, DDS/PhD, Department of Dental Surgery, Hôpital Maison Blanche, Reims University Hospital, UR BIOS (Biomaterials and Inflammation in Bone Site), Reims Champagne-Ardennes University, Reims, France

V**Arnaud VANJAK**

MD, PhD, Rheumatology Department, Lariboisière Hospital (AP-HP); Reference Center for Fibrous Dysplasia of Bone, BIOSCAR UMR 1132, Paris, France

Frédéric VELARD

PhD, HDR, Research Engineer, Head of a Research Subgroup of Bone loss: pathophysiology and therapeutic approaches at UR BIOS (Biomaterials and Inflammation in Bone Site), Reims Champagne-Ardennes University, Reims, France

W**Marjolaine WILLEMS**

MD, Medical Genetics, Head of Medical Genetics Department, Reference Center for Rare Bone Diseases, University Hospital of Montpellier, Institute for Neurosciences of Montpellier - INSERM U1051, Montpellier, France





12:00 - 13:20 Lunch break

OPENING CEREMONY

13:20 - 13:30 Agnès LINGLART (Le Kremlin-Bicêtre, France)

KEYNOTE LECTURE

Chairs: Valérie CORMIER-DAIRE (Paris, France), Adalbert RAIMANN (Vienna, Austria)

13:30 - 14:00 **Genetic disorders of the skeleton: a developmental approach**
Uwe KORNAK (Göttingen, Germany)

SESSION 1 - SHORT STATURE AND BONE GROWTH

Chairs: Valérie CORMIER-DAIRE (Paris, France), Adalbert RAIMANN (Vienna, Austria)

14:00 - 14:30 **Genetics of short stature and mild skeletal anomalies**
Karen E. HEATH (Madrid, Spain)

14:30 - 15:00 **The mouse model of hypochondroplasia**
Laurence LEGEAI-MALLET (Paris, France)

15:00 - 15:30 Short oral communications
Prevention of height deficit by burosumab in toddlers affected by XLH
Alexandra ERTL (Le Kremlin-Bicêtre, France)

Lower limb lengthening outcomes in achondroplasia: timing of surgery and impact on height and functionality
Inês ALVES (Évora, Portugal)

15:30 - 16:00 **Interactive clinical cases by attendees and faculty**
Valérie CORMIER-DAIRE (Paris, France), Thomas EDOUARD (Toulouse, France),
Marjolaine WILLEMS (Montpellier, France)

16:00 - 16:30 Coffee break and Posters





SESSION 2 - LESSONS FROM THE GROWTH PLATE AND TOOLS

Chairs: Justine BACCHETTA (Lyon, France), Corinna GRASEMANN (Bochum, Germany)

16:30 - 17:00 Characterization of bone mineralization in rare diseases

Nadja FRATZL-ZELMAN (Vienna, Austria)

17:00 - 17:30 The input of new technologies for bone tissues

Thomas Levin Geiser ANDERSEN (Odense, Denmark)

17:30 - 18:00 Short oral communications

Continuous subcutaneous parathyroid hormone (PTH 1-34) infusion (CSPI) in children and young people with severe autosomal dominant hypocalcaemia type 1 (ADH1); long term follow-up

Aikaterini PEROGIANNAKI (London, UK)

Genetic insights into short stature: an evaluation of clinical, hormonal, and genetic parameters in a real world paediatric cohort

Virginia ROSSI (London, UK)

18:00 End of day 1 sessions

20:00 Dinner





SESSION 3 – TEETH AS A MODEL TO STUDY BONE PATHOLOGY

Chairs: Catherine CHAUSSAIN (Paris, France), Martin BIOSSE DUPLAN (Paris, France)

08:30 - 09:00	Autoimmune amelogenesis imperfecta in APS1 Jakub ABRAMSON (Rehovot, Israel)
09:00 - 09:30	Rare disorders and impaired tooth mineralization Brian FOSTER (Ohio, USA)
09:30 - 10:00	Therapeutic strategies of X-linked hypophosphatemia Claire BARDET (Paris, France)
10:00 - 10:30	Short oral communications ENAMEL phenotype of mouse models of familial hypocalciuric hypercalcemia types 2 and 3 Anne-Laure Leyla LAKHEL (Paris, France) Identification of amelogenesis imperfecta in patients with hypercalcemia due to GNA11 loss-of function mutations Nicolas OBTEL (Paris, France)
10:30 - 11:00	Coffee break and Posters

SESSION 4 – WHAT'S NEW IN VITAMIN D?

Chairs: Agnès LINGLART (Le Kremlin-Bicêtre, France), Raja PADIDELA (Manchester, UK)

11:00 - 11:30	Manipulation of the VDR Gilles LAVERNY (Strasbourg, France)
11:30 - 12:00	Nutritional rickets in the world Wolfgang HÖGLER (Linz, Austria)
12:00 - 12:30	Short oral communications Assessment of placental Vitamin D transport in a UK cohort enriched for pregnancies with suboptimal fetal growth Reena PERCHARD (Manchester, UK) Empiric Vitamin D supplementation: a potential strategy to prevent type 2 diabetes? Salome BATSASHVILI (Tbilisi, Georgia)
12:30 - 13:00	Interactive clinical cases by attendees and faculty Justine BACCHETTA (Lyon, France), Agnès LINGLART (Le Kremlin-Bicêtre, France), Arnaud MOLIN (Caen, France)
13:00 - 14:00	Lunch break and Posters





SESSION 5 – FIBROUS DYSPLASIA OF BONE

Chairs: Roland CHAPURLAT (Lyon, France), Frédéric VELARD (Reims, France)

14:00 - 14:30 Lessons from the animal models

Mara RIMINUCCI (Rome, Italy)

14:30 - 15:00 Clinical landscape from the registries

Natasha APPELMAN-DIJKSTRA (Leiden, The Netherlands)

15:00 - 15:30 Short oral communications

Implication of autotaxine in oral fibrous dysplasia of bone

Johan SERGHERAERT (Reims, France)

Progression of spinal fibrous dysplasia lesions in adults: a 10-year follow-up study

Arnaud VANJAK (Paris, France)

15:30 - 16:00 Coffee break and Posters

SESSION 6 – ROUND TABLE ON RECENT GUIDELINES AND PERSPECTIVES PATIENTS

Chair: Agnès LINGLART (Le Kremlin-Bicêtre, France)

16:00 - 17:00 Achondroplasia

Valérie CORMIER-DAIRE (Paris, France)

X-linked hypophosphatemia (XLH)

Dieter HAFFNER (Hannover, Germany)

Patient Representative Group – Association RVRH-XLH

Frédéric GAULOIS (Suresnes, France)

CLOSING PLENARY CONFERENCE

Chair: Hervé KEMPF (Nancy, France)

17:00 - 17:30 The genetics of hereditary disorders of ectopic calcification

Frank RUTSCH (Münster, Germany)

17:30 Closure of the meeting and departures



ESPE-OSCAR
PARIS 2025



SCAN THE QR CODE TO FIND ALL THE INFORMATION ABOUT
THE SCIENCE SYMPOSIUM ESPE-OSCAR 2025

www.espe-oscar-science-symposium-2025.org

CONTACTS

ESPE

EUROPEAN SOCIETY OF PEDIATRIC ENDOCRINOLOGY

<https://www.eurospe.org/>

OSCAR FILIÈRE SANTÉ MALADIES RARES

DE L'OS, DU CALCIUM ET DU CARTILAGE

contact@filiere-oscar.fr - www.filiere-oscar.fr

OSCAR TEAM

Sara BREDACHE

Elisabeth CELESTIN

Yahya DEBZA

Meryem SARI HASSOUN