

### TUESDAY 17 NOVEMBER

15:00-16:00

#### ICCBH New Investigator Networking Group meeting

Organisers: Adalbert Raimann (*Vienna, Austria*) & Marie-Eve Robinson (*Ottawa, Canada*)

Paediatric Bone Health: navigating the path to a meaningful career

Laura Bachrach (*Stanford, USA*)

Followed by discussion and networking

23:00-02:00

#### OIF Young Investigator Symposium

(separate platform, accessed through the OIF website)

### WEDNESDAY 18 NOVEMBER

14:55-15:00

#### Welcome

Frank Rauch (*Montreal, Canada*) ICCBH Steering Group Chair

15:00-16:00

#### Clinical care in osteogenesis imperfecta

IS1 The impact of osteogenesis imperfecta  
Argerie Tsimicalis (*Montreal, Canada*)


IS2 Bisphosphonates in osteogenesis imperfecta: evidence and gaps in knowledge  
Bente Langdahl (*Aarhus, Denmark*)


#### 16:00-16:15 POSTERS

16:15-17:15

#### Oral communications 1

OC1 A multicenter study of intramedullary rodding in osteogenesis imperfecta  
Mercedes Rodriguez-Celin (*Chicago, USA*)

 OC2 The zebrafish p3h1<sup>-/-</sup> models osteogenesis imperfecta type VIII and represents a tool for drug screening approaches  
Valentina Daponte (*Pavia, Italy*)

 OC3 New Ifitm5 S42L mouse model for atypical type VI osteogenesis imperfecta recapitulates patient phenotype  
Gali Guterman-Ram (*Baltimore, USA*)

OC4 Bone tissue and osteoblasts from X-linked type XVIII OI with defects in regulated membrane proteolysis have distinct features  
Allahdad Zarei (*Columbia, USA*)

OC5 Genetic inactivation of sclerostin in a mouse model of severe osteogenesis imperfecta  
Iris Boraschi (*Montreal, Canada*)

#### 17:15-17:30 POSTERS

17:30-18:30

#### Workshop: Patient involvement in research

Moderators: Tracy Hart (OIF)/Ingunn Westerheim (OIFE)

Short guide to patient partnerships in rare disease research projects

Virginie Bros-Facer (*Paris, France*)

What I learned from involving patients in my research

Lars Folkestad (*Odense, Denmark*)

Patient involvement - A box to tick off or making a real difference?

Ingunn Westerheim (*Oslo, Norway*)

18:30-19:00

#### Osteogenesis imperfecta basic science

IS3 Effect of sclerostin inhibition in osteogenesis imperfecta mouse models  
Ken Kozloff (*Ann Arbor, USA*)

19:00-20:00

#### Meet the expert sessions

Osteogenesis imperfecta outcome measures

Claire Hill (*Sheffield, UK*)

With team members Davina Ford, Caroline Marr and Lisa Mills

Muscle-bone unit in osteogenesis imperfecta mouse models

Charlotte Phillips (*Columbia, USA*)

20:00-21:00

#### Meet the speakers and networking



Tweeting about the meeting?

Please use hashtag **#iccbhonline**

## THURSDAY 19 NOVEMBER

12:45-14:45

### Pediatric Hypophosphatasia: A Virtual Update for 2020

ORGANISED BY SOFT BONES

Overview

Michael Whyte (*St Louis, USA*)

Genetic etiology

Steven Mumm (*St Louis, USA*)

In utero and perinatal considerations

Deborah Krakow (*Los Angeles, USA*)

Treatment

Jill Simmons (*Nashville, USA*)

15:00-16:00

### Genetic causes of osteoporosis

IS4 Monogenic osteoporosis

Outi Mäkitie (*Helsinki, Finland*)


IS5 Polygenic osteoporosis

Brent Richards (*Montreal, Canada*)

### 16:00-16:15 ATTENDED POSTERS (ODD NUMBERS)

16:15-17:15

### Oral communications 2

 OC6 Evaluation of epigenetic regulation of osteogenesis imperfecta severity with circulating microRNAs: the miROI study

Alexandre Mercier (*Lyon, France*)

OC7 The proteomic signature of osteoblast-derived extracellular vesicles in osteogenesis imperfecta caused by COL1A1 mutations

Hadil Al-Jallad (*Montreal, Canada*)

OC8 Antagonism between PEDF pathway and TGF- $\beta$  pathway is implicated in type VI OI pathogenesis

Heeseog Kang (*Bethesda, USA*)

OC9 Bone cell functions in peptidylprolyl cis-trans isomerase B (PPIB) knock-out mouse model for type IX osteogenesis imperfecta (OI) are distinct from classical dominant OI

Ying Liu (*Rockville, USA*)

OC10 Establishing a critical-sized segmental defect in a mouse model of osteogenesis imperfecta

Kyle Kavaseri (*Montreal, Canada*)

### 17:15-17:30 ATTENDED POSTERS (ODD NUMBERS)

17:30-18:15

### Moderated posters: P1 – P9

### 18:15-18:30 ATTENDED POSTERS (ODD NUMBERS)

18:30-19:00

### Bone fragility basic science

IS6 NOTCH signalling and bone fragility

Ernesto Canalis (*Farmington, USA*)

19:00-20:00

### Meet the expert sessions

Bone fragility findings through various imaging modalities

Amaka Offiah (*Sheffield, UK*)

Altered intracellular homeostasis in bone fragility

Katarzyna Piróg (*Newcastle upon Tyne, UK*)

20:00-21:00

### Meet the speakers and networking

## FRIDAY 20 NOVEMBER

15:00-16:00

### Osteoporosis in serious conditions

IS7 Management of impaired bone health in children and adolescents with eating disorders

Madhu Misra (*Boston, USA*)

IS8 Bone fragility and morbidity in Duchenne muscular dystrophy: management strategies and unmet needs

Leanne Ward (*Ottawa, Canada*)

### 16:00-16:15 ATTENDED POSTERS (EVEN NUMBERS)

16:15-17:15

### Oral communications 3

OC11 In-utero denosumab exposure throughout pregnancy: a case report

Ravit Regev (*Toronto, Canada*)

OC12 A data-driven analysis of bone structure in osteogenesis imperfecta using HR-pQCT images and machine learning

Pouyan Asgharzadeh (*Montreal, Canada*)

OC13 Recombinant human ENPP1-Fc prevents ectopic tissue calcification and restores growth in ENPP1 deficient mice

Zhiliang Cheng (*Boston, USA*)

OC14 Efficacy and safety of burosumab in children and adolescents with X-linked hypophosphatemia: a prospective cohort

Volha Zhukouskaya (*Le Kremlin Bicêtre, France*)

OC15 In vitro safety of bisphosphonate and PTH treatment during childhood acute lymphoblastic leukemia therapy

Demi de Winter (*Rotterdam, Netherlands*)

### 17:15-17:30 ATTENDED POSTERS (EVEN NUMBERS)

17:30-18:15

### Moderated posters: P10 – P18

### 18:15-18:30 ATTENDED POSTERS (EVEN NUMBERS)

18:30-19:30

### Clinical conundrums: interactive cases

Where has all the phosphate gone?

Gabriele Häusler/Adalbert Raimann (*Vienna, Austria*)

19:30-20:00

### Management of secondary osteoporosis

IS9 Management of secondary osteoporosis in children

David Weber (*Philadelphia, USA*)

20:00-20:05

### Close

Frank Rauch (*Montreal, Canada*) ICCBH Steering Group Chair

20:05-21:00

### Meet the speakers and networking



Tweeting about the meeting?

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- P1** Beneficial effect of burosumab on hyperparathyroidism in children with X-linked hypophosphatemia  
*Volha Zhukouskaya (Le Kremlin-Bicêtre, France)*
- P2** Effects of treatment with bone-targeted prostaglandin E2 receptor 4 agonist C3 (mes-1007) in a mouse model of severe osteogenesis imperfecta  
*Iris Boraschi (Montreal, Canada)*
- P3** Bone quality during skeletal development: structural and compositional adaptations on the micro- and nanoscale  
*Kilian Stockhausen (Hamburg, Germany)*
- P4** Studies of OI Patient and Murine Osteoblasts to Investigate Phenotypic Variability of Dominant Osteogenesis Imperfecta  
*Milena Jovanovic (Bethesda, United States)*
- P5** Non-collagen mutations in children with OI - a national perspective  
*Patrick Thorney (Sheffield, UK)*
- P6** Abnormal energy metabolism in a mouse model for craniometaphyseal dysplasia  
*Shyam Kishor Sah (Farmington, United States)*
- P7** Developing and validating clinical risk prediction models for symptomatic fractures and low bone mineral density in pediatric acute lymphoblastic leukemia  
*Emma Jacobine Verwaaijen (Utrecht, Netherlands)*
- P8** Analysis of growth pattern in acrodysostosis  
*Diana-Alexandra Ertl (Vienna, Austria)*
- P9** Characterization and functional analysis of the adipose tissue-derived stromal vascular fraction of pediatric patients with osteogenesis imperfecta  
*Josephine T Tauer (Montreal, Canada)*
- P10** Collagen retention perturbs homeostasis in Osteogenesis Imperfecta cells and can be rescued by a chemical chaperone  
*Nadia Garibaldi (Pavia, Italy)*
- P11** 4-phenylbutyric acid likely improves the quality of extracellular matrix and promotes mineralization in patients with osteogenesis imperfecta-derived cells.  
*Shinji Takeyari (Osaka, Japan)*
- P12** Osteoporosis treatment with GIP and GLP-2 dual-agonists based on their synergistic actions in humans - a novel therapeutic principle in children and premenopausal women suffering from bone fragility  
*Maria Buur Nordskov Gabe (Copenhagen, Denmark)*
- P13** Matched angle image registration improves short-term precision of bone strength estimated from HR-pQCT scans of adults with osteogenesis imperfecta  
*Seyedmahdi Hosseinitabatabaei (Montreal, Canada)*
- P14** Male but not female mice, with a dominant severe form of osteogenesis imperfecta, are protected against high-fat diet-induced weight gain  
*Josephine T Tauer (Montreal, Canada)*
- P15** Skeletal response to mechanical stimulation in children with osteogenesis imperfecta  
*Sivagamy Sithambaram (Manchester, United Kingdom)*
- P16** Craniocervical abnormalities in osteogenesis imperfecta Type V  
*Karissa Ludwig (Montreal, Canada)*
- P17** Impact of the COVID-19 pandemic on patients with osteogenesis imperfecta  
*Malinda Wu (Atlanta, United States)*
- P18** Recombinant human parathyroid hormone (1-84) replacement therapy in childhood hypoparathyroidism: a case report  
*Elisabeth Laurer (Linz, Austria)*
- P19** Combined zoledronic acid and growth hormone treatment in osteogenesis imperfecta  
*Yael Levy-Shraga (Ramat Gan, Israel)*
- P20** Body composition and bone mineral density in craniopharyngioma patients: a longitudinal study over 10 years  
*Selveta S Van Santen (Rotterdam, Netherlands)*
- P21** Optimizing care through the mobility clinic for Filipino patients with osteogenesis imperfecta  
*Ebner Bon Maceda (Manila, Philippines)*
- P22** Monthly intravenous alendronate treatment can maintain bone strength in osteogenesis imperfecta patients following cyclical pamidronate treatment.  
*Daisuke Harada (Osaka, Japan)*
- P23** What skeleton imaging modality is best for assessing bone health in children and young adults?  
*Heba Shalof (Sheffield, United Kingdom)*
- P24** What is the correlation between skeletal burden score and functional outcomes in fibrous dysplasia?  
*Arwa Alhulwah (Sheffield, United Kingdom)*
- P25** A case of Vitamin D-dependent rickets type I, presenting with failure to thrive and marked hypotonia.  
*Artemis Doulgeraki (Athens, Greece)*
- P26** Infant with hepatoblastoma and rickets. Is there a link?  
*Artemis Doulgeraki (Athens, Greece)*
- P27** Can hypokalaemia be a complication of pseudohypoparathyroidism?  
*Amanda Peacock (Sheffield, United Kingdom)*
- P28** Juvenile Paget's disease: case report of a rare condition  
*Ana Rita Prata (Coimbra, Portugal)*
- P29** Modification of the osteogenesis imperfecta quality of life scale - pediatric version (OIQoL-P) and development of a parent-report version  
*Claire Hill (Sheffield, UK)*
- P30** Osteogenesis imperfecta cannot be excluded by fracture type  
*Amy Bobyn (Edmonton, Canada)*

- P31** Osteogenesis imperfecta type 1 in a patient with pseudo-vitamin D deficiency rickets: when a rare genetic bone disease hides another  
*Kim Phung (Montreal, Canada)*
- P32** Novel pathogenic heterozygous SLC34A1 mutation in a patient with idiopathic infantile hypercalcemia  
*Kim Phung (Montreal, Canada)*
- P33** The use of phospho-soda solution in management of an 11-year-old boy with hypophosphatemic rickets and pre-existing nephrocalcinosis  
*Ghaisani Fadiana (Jakarta Pusat, Indonesia)*
- P34** PHEX mutation in a Malaysian hypophosphatemic rickets patient  
*Tzer Hwu Ting (Serdang, Malaysia)*
- P35** Late presentation of vitamin D dependent rickets type-2 (VDDR II) following hepatic transplantation for progressive familial intrahepatic cholestasis (PFIC)  
*Sumudu Nimali Seneviratne (Colombo, Sri Lanka)*
- P36** Case history: A premature infant with multiple fractures  
*Sumudu Seneviratne (Rajagiriya, Sri Lanka)*
- P37** Hepatic osteodystrophy: an under-recognised complication of chronic liver disease  
*Sumudu Seneviratne (Rajagiriya, Sri Lanka)*
- P38** Children with osteogenesis imperfecta sustain patella injuries relatively commonly and undergo variable management.  
*Hannah Bassett (Bristol, United Kingdom)*
- P39** Health-related quality of life in paediatric patients with osteogenesis imperfecta  
*Adalbert Raimann (Vienna, Austria)*
- P40** Growth hormone treatment improves final height in children affected by X-linked hypophosphatemia  
*Volha Zhukouskaya (Paris, France)*
- P41** Craniofacial and dental phenotype of two girls with osteogenesis imperfecta due to mutations in CRTAP  
*Juliana Marulanda (Montreal, Canada)*
- P42** Fibrodysplasia ossificans progressiva masquerading as nodular fasciitis  
*Tiffany Sin-ting Lai (Hong Kong, Hong Kong)*
- P43** Establishing the first pan-European Registry for Rare Bone and Mineral Disorders: EuRR-Bone  
*Marina Mordenti (Bologna, Italy)*
- P44** Experiences of delivering virtual therapy groups for children with osteogenesis imperfecta during the Covid-19 pandemic  
*Lisa Mills (Bristol, United Kingdom)*
- P45** Inactivation of the Gorab gene does not alter the mechanoresponse  
*Beatrice Steyn (Montreal, Canada)*
- P46** Bone mineral density and content for forearm in children athletes; gymnastics vs swimmers  
*Magdy Abouzeid (Alexandria, Egypt)*
- P47** Calvarial doughnut lesions with bone fragility: a story of 6 generations in a French-Canadian family  
*Shuaa Basalom (Charlottetown, Canada)*
- P48** Post-transplantation refractory hypercalcemia in osteopetrosis caused by a novel mutation in TNFRSF11A (RANK)  
*Marie-Eve Robinson (Ottawa, Canada)*
- P49** Genetic heterogeneity of osteogenesis imperfecta in a large Turkish cohort  
*Sare Betul Kaygusuz (Istanbul, Turkey)*
- P50** Clinical themes from a contemporary series of children with Type XI osteogenesis imperfecta associated with FKBP10  
*Christine P Burren (Bristol, United Kingdom)*
- P51** Impacted permanent dentition in an adolescent with osteogenesis imperfecta type XII  
*Joanna Yuet-ling Tung (Hong Kong, Hong Kong)*
- P52** Clinical outcome of patients with osteogenesis imperfecta on intravenous pamidronate treatment at the Philippine General Hospital from 2010-2018  
*Cheryll Magbanua-Calalo (Quezon City, Philippines)*
- P53** Acute lymphoblastic leukemia in a child with osteogenesis imperfecta type V  
*Renee Anne Karmela L Feliciano (Metro Manila, Philippines)*
- P54** Panostotic fibrous dysplasia in newly diagnosed McCune Albright syndrome: an Indonesia experience  
*Frida Soesanti (Jakarta, Indonesia)*
- P55** Lithuanian boy with osteogenesis imperfecta in combination with pseudoachondroplasia  
*Rasa Traberg (Kaunas, Lithuania)*
- P56** Known COL1A1 variant, expanded phenotype?  
*Catarina S. Rosas (Coimbra, Portugal)*
- P57** Osteogenesis imperfecta in mother - baby dyads – what a difference a bisphosphonate makes?  
*Sarah-Jane O'Riordan (Dublin, Ireland)*
- P58** Chronic and refractory hypocalcemia in an infant with autosomal recessive polycystic kidney disease  
*Patricia Diaz Escagedo (Montreal, Canada)*