Scientific Programme

Times shown are Central European Time
(eg 15:00 Paris = 14:00 London, 09:00 Washington DC, 06:00 San Francisco, 23:00 Sydney)

ICCBH VIRTUAL FORUM
Bone Fragility Disorders in Children
18-20 November 2020

TUESDAY 18 NOVEMBER

15:00-16:00
ICCBH New Investigator Networking Group meeting

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

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-/- 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 project
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
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)*

Muscle-bone unit in osteogenesis imperfecta mouse models
Charlotte Phillips *(Columbia, USA)*

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THURSDAY 19 NOVEMBER

15:00-16:00
Genetic causes of osteoporosis
Monogenic osteoporosis
Outi Mäkitie *(Helsinki, Finland)*

Polygenic osteoporosis
Brent Richards *(Montreal, Canada)*

16:00-16:15 POSTERS

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-β 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 POSTERS

17:30-18:15
Moderated posters: P1 – P9

18:30-19:00
Bone fragility basic science
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)*

FRIDAY 20 NOVEMBER

15:00-16:00
Osteoporosis in serious conditions
Management of impaired bone health in children and adolescents with eating disorders
Madhu Misra *(Boston, USA)*

Fractures in Duchenne muscular dystrophy
Leanne Ward *(Ottawa, Canada)*

16:00-16:15 POSTERS

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 POSTERS

17:30-18:15
Moderated posters: P10 – P18

18:30-19:30
Clinical conundrums: interactive case session
Where has all the phosphate gone?
Gabriele Häusler/Adalbert Raimann *(Vienna, Austria)*

19:30-20:00
Management of secondary osteoporosis
Management of secondary osteoporosis in children
David Weber *(Philadelphia, USA)*

20:00-20:05
Close
Frank Rauch *(Montreal, Canada)* ICCBH Steering Group Chair

Poster List

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 adaptions 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 3D image registration improves short-term in-vivo precision of bone strength in high resolution peripheral quantitative CT scans from adults with osteogenesis imperfecta in the presence of motion artefacts
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 (Leeds, United Kingdom)

P28 Juvenile Paget's disease: case report of a rare condition
Ana Rita Prata (Coimbra, Portugal)

Poster List

P29 Modification of the osteogenesis imperfecta quality of life scale - pediatric version (OIQoL-P) and development of a parent-report version
Elizabeth Gibbons (Chicago, United States)

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)