

Bone development

- P1 How to grow a synovial joint: the cell-level activities underlying prenatal joint morphogenesis

 Niamh Nowlan (Dublin, Ireland)
- P2 Observational study of fetal foramen magnum and foramen magnum stenosis development in Achondroplasia

 Rhoda Akilapa (London, UK)
- P3 Variation in skeletal robustness is an important determinant of bone strength in youth and young adults in both males and females

 Vanessa Yingling (Hayward, USA)
- P4 ABSTRACT WITHDRAWN
- P5 Variations of the trabecular bone microarchitecture through the period of locomotor behaviour development of a late 19th century post-industrial Italian population.

 Antony Colombo (Pessac cedex, France)
- Comparison of cortical hone outcomes in
- P6 Comparison of cortical bone outcomes in the metaphysis and diaphysis during growth Kyla Kent (Palo Alto, USA)
- P7 The applicability and agreement between two methods of bone age assessment in Zimbabwean children and adolescents

 Farirayi Kowo-Nyakoko (Harare, Zimbabwe)
- P8 Predictors of skeletal maturity in Zimbabwean children and adolescents

 Farirayi Kowo-Nyakoko (Harare, Zimbabwe)
- P9 Fun with 50: testing the legend of height prediction at age two

 Dana Duren (Columbia, USA)

POSTER KEY

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P26-P36	Bone metabolism
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Bone fragility

- P10 Elevated bone turnover markers predict increased bone mineral density in adolescents with congenital adrenal hyperplasia

 Pattara Wiromrat (Khon Kaen, Thailand)
- P11 Bone health in patients with type 1 diabetes and progressive eGFR decline from adolescence into young adulthood

 Funmbi Babalola (Toronto, Canada)
- P12 Does body mass index influence the risk of vertebral fractures in children with osteogenesis imperfecta?

 Sameera Auckburally (Manchester, UK)
- P13 Spontaneous reshaping of vertebral fractures in primary and secondary osteoporosis

 Rodrigo Montero-Lopez (Linz, Austria)
- P14 Pubertal growth in osteogenesis imperfecta caused by pathogenic variants in COL1A1/COL1A2 Marie-Eve Robinson (Ottawa, Canada)
- P15 Burden of steroid use in the treatment of Duchenne Muscular Dystrophy: Findings from a targeted literature review, online survey and qualitative research as part of Project HERCULES Caleb Hariri (Shotts, UK)
- P16 Histomorphometric answers to vertebral changes in sickle cell disease.

 Sophia Sakka (London, UK)
- P17 Platelet-derived growth factor type BB is decreased in boys with Duchenne Muscular Dystrophy on chronic glucocorticoids without history of fracture

 Julia See (Miami, USA)
- P18 SMAD3 mutation in LDS3 causes bone fragility by impairing the TGF-β pathway and enhancing osteoclastogenesis

 Ahmed El-Gazzar (Linz, Austria)
- P19 Risk of chronic diseases among children with a significant fracture history

 Yael Levy Shraga (Ramat Gan, Israel)
- P20 Use of zoledronate in children with sickle cell disease: a single centre experience Sophia Sakka (London, UK)
- P21 Use of oral risedronate in children with osteoporosis during Covid pandemic: Evelina experience

 Jaya Sujatha Gopal-Kothandapani (London, UK)
- P22 Horizonal gaze palsy with progressive scoliosis secondary to ROBO3 gene variants associated with osteogenesis imperfecta type I Belinda Crowe (London, UK)



- P23 Catastrophic vertebral fracture cascade in boys with Duchenne muscular dystrophy on long-term glucocorticoids
 - Nicola Crabtree (Birmingham, UK)
- P24 KDELR2-related osteogenesis Imperfecta (OI21): clinical and radiological features in two siblings with novel compound heterozygous intragenic deletions
 - Catherine DeVile (London, UK)
- P25 Multiple vertebral fractures: Was the cause hiding in plain sight: Lifestyle?

 Toby Candler (Bristol, UK)

Bone metabolism

- P26 Hereditary hypophosphatemic rickets and osteomalacia an uncommon cause of metabolic bone disease

 Shivani Shivani (Bathinda, India)
- P27 The results of pharmaceutical therapy for structural disorders of bone in patients with fibrous dysplasia Alexander Skuratov (Kyiv, Ukraine)
- P28 Novel PTH gene mutations causing isolated hypoparathyroidism

 Michael Levine (Philadelphia, USA)
- P29 The peculiarity of the bone metabolism in children with Wilson's disease

 Nataliya Balatska (Kyiv, Ukraine)
- P30 Skeletal remodelling and resolution of recurrent severe hypercalcaemia following withdrawal of prolonged denosumab after haematopoietic stem cell transplant for TNFRSF11A osteopetrosis Christine Burren (Bristol, UK)
- P31 Osteocalcin drives metabolic adaptation in a mouse model of severe dominant Osteogenesis Imperfecta.

 Josephine Tauer (Montreal, Canada)
- P32 Coeliac disease presenting with vertebral fractures in a child with multiple epiphyseal dysplasia Stephanie Borg (Sheffield, UK)
- P33 Adiponectin has a pro-resorption role on bone in childhood-onset brain tumors

 Natascia di lorgi (Genoa, Italy)
- P34 Spontaneous remission of hypophosphataemic rickets in a child.

 Nick Shaw (Birmingham, UK)
- P35 Transient hyperphosphatasemia in a child with autism spectrum disorder

 Stepan Kutilek (Klatovy, Czech Republic)
- P36 Serum osteocalcin levels in children with steroidresistant nephrotic syndrome Aashima Dabas (New Delhi, India)

Diagnostics and imaging

- P37 Reduced responsivity of the skull to stimuli associated with bone mineralisation at other skeletal sites in children

 Rebecca Moon (Southampton, UK)
- P38 Radiology reporting of paediatric osteoporotic vertebral fractures in Duchenne muscular dystrophy and potential impact on clinical management

 Hannah Martin (Glasgow, Scotland)
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 Jananie Suntharesan (Liverpool, UK)
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 Nandhini Perumal (Manchester, UK)
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 Jean De Schepper (Brussels, Belgium)
- P43 What's in a name? Prevalence of metaphyseal fractures in children with osteogenesis imperfecta in the first two years of life

 Ella Riley (Sheffield, England)
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 Carla Caffarelli (Siena, Italy)
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 Ella Riley (Sheffield, England)
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 Ammar Abusultan (Al Khobar, Saudi Arabia)
- P47 MRI investigation of vertebral changes in children with sickle cell disease

 Jaya Sujatha Gopal-Kothandapani (London, UK)
- P48 Joint effusions as a radiographic feature of complex chronic recurrent multifocal osteomyelitis (CRMO)

 loanna Tsigkouli (Stanmore, UK)



P49 A reference range for plasma levels of inorganic pyrophosphate in children

Eva Bernhard (Münster, Germany)

Genetics

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 Karissa Ludwig (Montreal, Canada)
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 Thomas Borge (Espoo, Finland)
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 Alistair Calder (London, UK)
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- P55 A de novo deleterious PHEX variant with no evidence of X-linked hypophosphatemia

 Michelle Kayser (New Haven, CT, USA)

Medical therapies

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 Matthew Benson (Jacksonville, USA)
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 Raja Padidela (Manchester, UK)
- P60 Growth pattern in children with X-linked hypophosphatemia treated with burosumab and growth hormone

 Diana-Alexandra Ertl (Vienna, Austria)

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- P62 An appraisal of a spine screening programme for vertebral fracture and its management in Duchenne Muscular Dystrophy:ls it time to reconsider threshold for treatment with bisphosphonate?

 Hannah Martin (Glasgow, Scotland)
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 Avivit Brener (Tel Aviv, Israel)
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 Benjamin Jacobs (Stanmore, UK)



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 Emely Loscalzo (Baltimore, US)
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 Jeremy Allgrove (London, UK)
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 Hamilton Cassinelli (CABA, Argentina)
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 Kebashni Thandrayen (Johannesburg, South Africa)
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 Vivian Szymczuk (Bethesda, USA)

Mineral homeostasis and vitamin D

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 Rebecca Moon (Southampton, UK)
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 Helena Hauta-alus (Helsinki, Finland)
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 Aashima Dabas (New Delhi, India)
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 Jarod Wong (Glasgow, UK)
- P85 Barakat syndrome -description of two cases Stepan Kutilek (Klatovy, Czech Republic)

- P86 Clinical course of hypoparathyroidism in children with autoimmune polyendocrine syndrome type-1 Saila Laakso (Helsinki, Finland)
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Muscle and bone

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 Soher Jayash (Edinburgh, UK)
- P89 A multi-disciplinary approach for bone-endocrine care in Duchenne Muscular Dystrophy: The Glasgow experience

 Jarod Wong (Glasgow, UK)
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 Jarod Wong (Glasgow, UK)
- P91 Physical impairments and activity limitations of childhood acute lymphoblastic leukemia survivors: A PETALE cohort study Annie Brochu (Montreal, Canada)
- P92 Delayed puberty is very common in boys with Duchenne muscular dystrophy on daily glucocorticoid

 Jarod Wong (Glasgow, UK)
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 Amish Chinoy (Manchester, UK)
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 Tandy Aye (Palo Alto, USA)
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Andrea Aul (Rochester, USA)



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health Adalbert Raimann (Vienna, Austria) Chariklia Pieridou (Coventry, UK)	1 110				
Adalbert Raimann (Vienna, Austria) P123 Gait deviations and burden of disease in children					
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with XLH

Alexandra Stauffer (Vienna, Austria)



- P124 ABSTRACT WITHDRAWN
- P125 Giant cell tumour in lumbar spine of an 11-year-old girl surgical and medical treatment with zoledronate and denusomab

 Signe Beck-Nielsen (Aarhus, Denmark)
- P126 Annual hearing screening in children with achondroplasia: results from the first 2 years in Glasgow

 Adam Watt (Glasgow, UK)
- P127 Weaker bones relative to body size in patients with thalassemia compared to healthy controls Vanessa Yingling (Hayward, USA)
- P128 Foramen magnum stenosis in infants with achondroplasia: investigating the natural history of cervicomedullary cord compression without signal change

 Connor Brett (London, UK)
- P129 Children with hypophosphatasia treated with asfotase alfa: interim analysis from the UK Cohort Raja Padidela (Manchester, UK)
- P130 Quantifying outcomes in fibrodysplasia ossificans progressiva (fop) by patient age: results from an international burden of illness survey

 Kim Croskery (Slough, UK)
- P131 Circulating collagen fragments as indicators for growth in children with achondroplasia

 Merete Ljungberg (Copenhagen, Denmark)
- P132 Reduced bone mineral accrual in pediatric patients with thalassemia Ellen Fung (Oakland, USA)
- P133 Parathyroid adenoma associated with Neurofibromatosis type 1 (NF1) A rare paediatric presentation.

 Jananie Suntharesan (Liverpool, UK)
- P134 Two heterozygous sequence variants of the CTSK gene in a girl with very small stature

 Milan Bayer (Prague, Czech Republic)
- P135 Identification of the CLCN7 mutation in children cause autosomal dominant osteopetrosis type II

 Voraluck Phatarakijnirund (Bangkok, Thailand)
- P136 Systematic review of muscle mass and muscle function in osteogenesis imperfecta

 Caleb Hariri (Shotts, UK)
- P137 Co-existence of osteogenesis imperfecta and hypophosphatasia pathological bone features in a 3-year-old boy with multiple bone fractures and mutations in COL1A1 and ALPL.

 Nadja Fratzl-Zelman (Vienna, Austria)
- P138 Early experience with vosoritide therapy in clinical practice

 Valérie Cormier-Daire (Paris, France)

- P139 Vosoritide for children with achondroplasia: growth velocity and pubertal milestones

 Melita Irving (London, UK)
- P140 Associations between height and health-related quality of life (HRQoL) and functional independence in children with achondroplasia Melita Irving (London, UK)
- P141 Should osteogenesis imperfecta be labeled as a low bone mass condition?

 Cathleen Raggio (New York, USA)
- P142 Skeletal disease acquisition in fibrous dysplasia:
 natural history and indicators of lesion progression
 in children
 Vivian Szymczuk (Bethesda, Maryland, USA)
- P143 Effects of a two-week intensive training program on motor skills and mobility in children and adolescents with musculoskeletal diseases Sandra Baumann (Linz, Austria)
- P144 Using virtual communication for rapid dissemination of COVID-19 information to individuals with osteogenesis imperfecta Laura Tosi (Washington, DC, USA)
- P145 Craniosynostosis in autosomal dominant hypophosphatemic rickets treated with ferrous sulfate

 Kelli Davis (Nashville, USA)
- P146 ABSTRACT WITHDRAWN
- P147 ACHieve Study: baseline characteristics of a multicenter observational study of children with achondroplasia

 Melita Irving (London, UK)
- P148 Baseline characteristics of the ACcomplisH phase 2 trial evaluating once-weekly TransCon CNP in children with achondroplasia

 Ciara McDonnell (Dublin, Ireland)
- P149 Serum phosphorus levels as a driver of skeletal morbidity in patients with fibrous dysplasia

 Zubeyir Hasan Gun (Bethesda, USA)
- P150 Do children aged 7-12 years old with achondroplasia have appropriate adaptations required to support independence in toileting?

 Kathryn Johnson (London, UK)
- P151 Are children with achondroplasia able to use the toilet independently on entry to primary school? Paul Watson (London, UK)
- P152 Evaluation of multidisciplinary care of children with chronic metabolic bone conditions at Royal Manchester Children's Hospital

 Nicola Panchbhaya (Manchester, UK)



- P153 Investigating the effect of a novel ASCC1 deletion mutation on human bone fragility

 Barbara Voraberger (Linz, Austria)
- P154 Functional testing of variants of uncertain significance in the ALPL gene: First results of the Hypophosphatasia gene variant consortium Ahmed El-Gazzar (Linz, Austria)
- P155 Hyperphosphatemic familial tumoral calcinosis: a case report

 Yael Levy Shrag (Ramat Gan, Israel)
- P156 Insights into endothelial and stromal microenvironment of osteosarcoma based on two cohorts of patients.

 Valérie Trichet (Nantes, France)
- P157 The diagnostic odyssey: lessons from Tin Soldiers and the search for undiagnosed individuals with fibrodysplasia ossificans progressiva (FOP)

 Christiaan Scott (Cape Town, South Africa)
- P158 The European Registries for Rare Bone and Mineral Conditions (EuRR-Bone): focusing on the core registry and disease specific modules

 Diana-Alexandra Ertl (Paris, France)
- P159 Treatment approach for hypophosphatasia with highly purified human mesenchymal stem cells "REC"

 Takeshi Taketani (Izumo-city, Japan)
- P160 The global Hypophosphatasia Gene Variant Database: Dedicated to deciphering variants Mariam Farman (Linz, Austria)
- P161 Relapse of giant cell granuloma on denosumab weaning as shown using semi-quantification of technetium Tc 99m hydroxydiphosphonate (Tc99m-HDP) uptake on SPECT imaging.

 Poonam Dharmaraj (Liverpool, Merseyside)
- P162 Engaging the osteogenesis imperfecta (OI) community in patient centered outcomes research Laura Tosi (Washington, USA)
- P163 Pseudohypoparathyroidism: focus on neonatal features, a retrospective analysis of a large cohort of patients

 Giulia Del Sindaco (Milan, Italy)
- P164 Annual hearing screening in children with osteogenesis imperfecta: results from first three years in Glasgow

 Emmett Lui (Glasgow, Scotland)
- P165 Phenotypic and genotype spectrum in a cohort of patients with Osteogenesis Imperfecta.

 Nalini M Selveindran (Selangor, Malaysia)
- P166 Mechanical regulation of bone healing is altered in mouse models of osteogenesis imperfecta

 David Bertrand (Montreal, Canada)

- P167 Primary hypertrophic osteoarthropathy: a case report with genetic and rheumatology evaluation and literature review

 Marija Mijovic (Belgrade, Serbia)
- P168 Hypochondroplasia: why are we still missing its diagnosis?

 Anna Allegri (Genova, Italy)
- P169 What is the impact of achondroplasia on function in children as measured by the Pediatric Evaluation of Disability Inventory- Computer Adaptive Test (PEDICAT)?

 Jill Massey (London, UK)
- P170 Multiple pathological fractures as a clinical sign of parathyroid adenoma in adolescent: a case report Frida Soesanti (Jakarta, Indonesia)
- P171 Craniofacial lesion progression in patients with fibrous dysplasia

 Jocelyn Taylor (Bethesda, USA)
- P172 Panostotic fibrous dysplasia in newly diagnosed McCune Albright syndrome: an Indonesia experience
 Frida Soesanti (Jakarta, Indonesia)
- P173 Insight into the bone dysplasia mechanism of CRTAP-null osteoblasts

 Aileen Barnes (Bethesda, USA)

Last minute abstracts

- LM4 A severe phenotype of Bruck syndrome caused by biallelic PLOD2 variant with additional phenotypic features of congenital cardiac disease and pulmonary haemorrhage. Craig Munns (Brisbane, Australia)
- LM5 Novel adapation to the SARC-F score to classify pediatric hemato-oncology patients with functional sarcopenia

 Emma Verwaaijen (Utrecht, Netherlands)
- LM6 Are we undertreating calcium deficiency in metabolic bone disease of prematurity? A case report

 Sirisha Kusuma Boddu (Hyderabad, India)
- LM7 Phenotyping fibrodysplasia ossificans progressiva using UK primary care electronic health records Orlando Buendia (London, UK)
- LM8 Late-onset severe primary hyperparathyroidism due to a novel homozygous CaSR mutation and the use of pamidronate in the post-operative management

 Sirisha Kusuma Boddu (Hyderabad, India)



- LM9 The IMPACT survey provides unique insights into the self-reported experiences of adolescents with osteogenesis imperfecta (OI) through a novel 95-participant dataset
 - Oliver Semler (Cologne, Germany)
- **LM10** Van Neck-Odelberg disease and its relevance to the painful hip in children

 Daniela Dyankova (London, UK)
- LM11 How we made an Impact The role of Patient
 Advocacy Groups in recruitment to surveys on rare
 conditions
 Ingunn Westerheim (Oslo, Norway)
- LM12 Characterization of pediatric inpatient admissions due to significant hypocalcemia with vitamin D deficiency

 Nadia Merchant (Washington, USA)
- LM13 Coned epiphyses with severe short stature in patient with abetalipoproteinemia

 Rashida Farhad Vasanwala (Singapore,

 Singapore)
- LM14 Establishing the prevalence of osteomalacia in Arab adolescents using nutritional biomarkers of bone health Nasser Al-Daghri (Riyadh, Saudi Arabia)
- LM15 Nontraumatic bilateral femoral and humerus fractures in an adolescent during a hypocalcemic seizure Sapna Nayak (Lucknow, India)
- **LM16** Atypical femoral fracture in type 1 osteogenesis imperfecta after discontinuation of bisphosphonate treatment

 **Rebecca Moon (Southampton, UK)*
- LM17 Assessment of health status of newborns with severe osteogenesis imperfecta – 21 years of observations Jakub Nowicki (Lodz, Poland)
- LM18 Vitamin D and osteocalcin levels among children with osteogenesis imperfecta – one-year observations Jakub Nowicki (Lodz, Poland)
- **LM19** Cross-sectional structure of cortical bone at the femoral midsection during early locomotor progression *Karen Swan (London, UK)*
- LM20 Ectonucleotide
 pyrophosphatase/phosphodiesterase 1 (ENPP1)
 deficiency appears to be associated with an
 evolving skeletal dysplasia in addition to ectopic
 calcification and rickets
 Deborah Wenkert (Thousand Oaks, USA)

- LM21 Ethanolic extract from C8 promotes fracture healing by reducing ROS level

 Kriti Sharma (Lucknow, India)
- LM22 ABSTRACT WITHDRAWN
- LM23 Premature epiphyseal fusion induced by a retinoic acid agonist in a young patient with fibrodysplasia ossificans progressiva

 Sigrún Hallgrímsdóttir (Solna, Sweden)
- LM24 Influence of chronic inflammation on bone mineral density in children with epidermolysis bullosa

 Natalia Balatska (Kyiv, Ukraine)
- LM25 Multicentric carpotarsal osteolysis syndrome.
 A Case Report
 Kelly Maury (Buenos Aires, Argentina)
- LM26 Metachondromatosis: Description of 5 new patients with a novel variant in PTPN11 Rhoda Akilapa (London, UK)
- LM27 Juvenile psammomatoid ossifying fibroma (JPOF) in the radius: A rare cause of tumour induced osteomalacia

 Benjamin Jacobs (Stanmore, UK)
- LM28 Bruck syndrome description of the first Polish infant with FKBP10 gene mutation

 Jakub Nowicki (Lodz, Poland)
- LM29 Vitamin D status and bone mineral density in children with allogeneic hematopoietic cell transplantation due to oncohematology

 Natalia Balatska (Kyiv, Ukraine)

Challenge the Expert Cases

A case of neonatal osteofibrous dysplasia with novel CDK12 and DDR2 mutations Ammar Abusultan (Al Khobar, Saudi Arabia)

Achondroplasia in a three-year-old male Siska Lubis (Medan, Indonesia)

Cutaneous-skeletal hypophosphatemic syndrome Margaret Zacharin (Melbourne, Australia)

Tumoral calcinosis

Margaret Zacharin (Melbourne, Australia)

OI/OI-like due to mutation in BMP1

Margaret Zacharin (Melbourne, Australia)