

14th International Conference on Osteogenesis Imperfecta
30 August – 2 September 2022, Sheffield, UK
Posters

P1

Significant modifiable risk factors for bone health in patients with osteogenesis imperfecta; the importance of lateral thinking.

Artemis Doulgeraki (Athens, Greece)

P2

Effect of bisphosphonate treatment on oral health status of children and adolescents with osteogenesis imperfecta

Artemis Doulgeraki (Athens, Greece)

P3

Identification of modifier genes underlying intra-familial phenotypic variability in zebrafish OI models using whole exome sequencing and linkage analysis

Tamara Jarayseh (Ghent, Belgium)

P4

Genetic analysis of Osteogenesis Imperfecta in Brazil

Temis Felix (Porto Alegre, Brazil)

P5

Characteristic modes of failure in single entry telescopic rods: a combined 2 centre experience

India Cox (Bristol, UK)

P6

Spine care in osteogenesis imperfecta from a distance: single center experience

Jeanne Franzone (Wilmington, USA)

P7

3D image registration marginally improves short-term in-vivo precision of high resolution peripheral quantitative CT images from adults with osteogenesis imperfecta compared to cross-sectional-area registration

Seyedmahdi Hosseinatabatabaei (Montreal, Quebec)

P8

Generation of Osteogenesis imperfecta stem cell bioresource to decipher disease mechanism and aid drug discovery

Chiara F Sander (Sheffield, UK)

P9

Oral health related quality of life in children with Osteogenesis Imperfecta of different ethnic backgrounds

Jasmine May Cachia Mintoff (London, UK)

P10

Bleeding assessment in 195 patients with Osteogenesis Imperfecta

Koert Gooijer (Zwolle, Netherlands)

P11

Mechano-regulation of bone formation and mineralization in healthy and type XI Osteogenesis Imperfecta patient-specific 3D bone organoids

Julia Griesbach (Zürich, Switzerland)

P12

Should Osteogenesis Imperfecta be Labeled as a Low Bone Mass Condition?

Cathleen Raggio (New York, USA)

P13

Surgical Correction of Skeletal Malocclusion in Osteogenesis Imperfecta

Joseph Napoli (Philadelphia, USA)

P14

Assessing the Safety and Efficacy of Tranexamic Acid Usage in Osteogenesis Imperfecta Patients Undergoing Femoral Rodding

Maegen Wallace (Omaha, USA)

P15

Therapeutic use of Indomethacin in Osteogenesis Imperfecta Type V

James Law (Birmingham, UK)

P16

Quality of life in Osteogenesis Imperfecta: evaluation of the content validity of the OIQoL questionnaire in an international cohort.

Claire Hill (Sheffield, UK)

P17

What are the perceived therapy needs of adults living with Osteogenesis Imperfecta? A focus group study.

Sophie Barlow (Stanmore, UK)

P18

A service evaluation examining the clinical application of the; Bleck, Brief Assessment of Motor Function Lower Limb and the Screening Tool for Everyday Mobility and Symptoms (STEM) in the functional assessment of children aged 6-18 years who have Osteogenesis Imperfecta

Carrie Marr (Sheffield, UK)

P19

Insight into the bone mechanism of CRTAP-null osteoblasts

Aileen Barnes (Bethesda, USA)

P20

Severe Obesity in Osteogenesis Imperfecta - What are the Treatment Options?

Jannie Dahl Hald (Aarhus, Denmark)

P21

Crispant analysis in zebrafish as a tool for rapid functional screening of disease-causing genes for Osteogenesis Imperfecta.

Sophie Debaenst (Ghent, Belgium)

P22

"O.I. wish orthopaedic surgeons had better strategies to help with..." - results of a patient and parent-based survey

Maegen Wallace (Omaha, USA)

P23

Physical activity of children with osteogenesis imperfecta during the period of immobilization (fractures) and during the «light» period (period of full functioning)

Nadezhda Epishina (St Petersburg, Russia)

P24

Investigating the transition from paediatric to adult services and the management of ongoing care of adults for osteogenesis imperfecta: A qualitative study

Chloe Morgan (Swansea, UK)

P25

Service evaluation reviewing the application of the Bayley Scale of Infant and Toddler development in children with severe, complex and atypical Osteogenesis Imperfecta.

Carrie Marr (Sheffield, UK)

P26

Cross-centre psychology collaboration on the adaptation of young person 'tree of life' narrative therapy groups to online during the COVID-19 pandemic, as part of the NHS England Osteogenesis Imperfecta National Service.

Rebecca Jones (Sheffield, UK)

P27

Cardiovascular characteristics and abnormalities in patients with Osteogenesis Imperfecta: a systematic review

Sara Verdonk (Amsterdam, The Netherlands)

P28

Molecular characterization of a newly reported MBTPS2 variant in a fetus affected with severe Osteogenesis Imperfecta

Giulio Marcionelli (Zurich, Switzerland)

P29

Studies of OI Patient and Murine Osteoblasts to Investigate Phenotypic Variability of Dominant Osteogenesis Imperfecta

Milena Jovanovic (Bethesda, USA)

P30

A new X-linked PLS3 gene variant identified in a patient with osteogenesis imperfecta

Petar Brlek (Zagreb, Croatia)

P31

Genotype-phenotype correlation and effects of bisphosphonates in rare forms of osteogenesis imperfecta : a retrospective study

Maëlle-Charlotte Charpié (Paris, France)

P32

Pulmonary Complications in Osteogenesis Imperfecta Type III

Hollis Chaney (Washington, DC, USA)

P33

Port-a-Cath Placement in Pediatric Patients with Osteogenesis Imperfecta Requiring Long-Term Vascular Access

Maegen Wallace (Omaha, USA)

P34

Intramedullary canal sclerosis as a result of prolonged bisphosphonate therapy in children with osteogenesis imperfecta type III and IV

Darko Antičević (Zagreb, Croatia)

P35

Tibial Rodding Challenges and Nailing Technique Choice in Children with Osteogenesis Imperfecta and Narrow Intramedullary Canals Using the "Telescoping Principles": A Narrative Review and Expert Opinion

Alice Tao Ran Guo (Montreal, Quebec, Canada)

P36

Bone microarchitecture in osteogenesis imperfecta assessed with high-resolution peripheral quantitative CT

Melissa Bevers (Venlo, The Netherlands)

P37

Early intervention program for children with OI. 5 years of experience of the Center for Inborn Pathology GMS Clinic

Natalia Belova (Moscow, Russia)

P38

Stiff hips in patients with Osteogenesis Imperfecta : a providential femoral neck fracture

Marine de Tienda (Paris, France)

P39

Tibia sliding elastic nailing technique in moderate-to-severe Osteogenesis Imperfecta :long-term outcomes

Marine de Tienda (Paris, France)

P40

Laboratory assessment in patients with the highest bleeding scores on the Self bleeding assessment tool out of 195 patients with Osteogenesis Imperfecta

Koert Gooijer (Zwolle, Netherlands)

P41

Understanding the Physical Well-being of Adults with Osteogenesis Imperfecta

Kathleen Montpetit (Montreal, Canada)

P42

Treatment response in Osteogenesis imperfecta - Two-year retrospective analysis of paediatric patients treated with bisphosphonates

Mirko Rehberg (Cologne, Germany)

P43

Relative larger Aortic Root Diameter in Osteogenesis Imperfecta Type 3 compared to type 1 and 4
Koert Gooijer (Zwolle, Netherlands)

P44

Tooth size in individuals with osteogenesis imperfecta
Henri Tuurala (Helsinki, Finland)

P45

Implementation of an Osteogenesis imperfecta patient registry to investigate clinical spectrum and genotype-phenotype correlations in OI
Timothée Ndarugendamwo (Zürich, Switzerland)

P46

The Fate of Bent Telescopic Rods in Children with Osteogenesis Imperfecta: Do All Bent Rods Need to be Revised?
David Fralinger (Wilmington, USA)

P47

Altered bone healing mechanoreponse exhibited by mouse models of osteogenesis imperfecta
David Bertrand (Montreal, Canada)

P48

Ophthalmological screening guidelines for individuals with osteogenesis imperfecta : a scoping review
Sarah Moussa (Montreal, Canada)

P49

Oral health related quality of life in adults with osteogenesis imperfecta, in Spain
Amira Elfituri (Madrid, Spain)

P50

Lack of psychosocial intervention assessment in OI: a scoping review
Marie-Catherine Mongenot (Westmount, Canada)

P51

Ifitm5/BRIL p.S42L murine model for atypical type VI OI has elevated serum alkaline phosphatase and disordered bone matrix but decreased bone cell function
Gali Guterman-Ram (Bethesda, USA & Haifa, Israel)

P52

Experience, problems and prospects of development of the program of diagnostics and treatment of osteogenesis imperfecta in children in the Republic of Kazakhstan.
Dossanova Assem (Nur-Sultan, Kazakhstan)

P53

Clinical management of the adult patient with Osteogenesis Imperfecta
Arjan Harsevoort (Zwolle, Netherlands)

P54

Patient Reported Experience Of Clinical Care Of Osteogenesis Imperfecta (OI) During The COVID-19 Pandemic

Coreen Kelday (Dundee, UK)

P55

A Dual centre transition clinic for young people with mild, moderate and severe Osteogenesis Imperfecta

Martyn Dudley (London, UK)

P56

Genetics of Rare Skeletal Disorders among Pakistani Consanguineous Families

Mehran Kausar (Gilgit, Pakistan)

P57

Prevalence and Hospital Admissions in Patients With Osteogenesis Imperfecta in The Netherlands: A Nationwide Registry Study

Silvia Storoni (Amsterdam, Netherlands)

P58

The European Registry for Rare Bone and Mineral Conditions (EuRR-Bone): Results of a Survey on Osteogenesis Imperfecta

Ana Luisa Priego Zurita (Leiden, Netherlands)

P59

How we made an Impact – The role of Patient Advocacy Groups in recruitment to surveys on rare conditions

Ingunn Westerheim (Oslo, Norway)

P60

Sleeping Compression - Lengthening Nail; Off-label use for treatment of non union then lengthening in Osteogenesis Imperfecta patient

Yasser Elbatrawy (Cairo, Egypt)

P61

Skull fractures, cervical spine fractures and intracranial injuries in children with Osteogenesis Imperfecta: a cohort study

Georgina Williams (London, UK)

P62

Long-Term Results of Initial and Reoperation Surgeries with Fassier-Duval Intramedullary Rods in Femurs and Tibias of Children with Osteogenesis Imperfecta

Maegen Wallace (Omaha, USA)

P63

Impact of pandemic on self-report measures for children with Osteogenesis Imperfecta(OI): a preliminary look

Maureen Donohoe (Wilmington, USA)

P64

'In-Out-In' K-wires sliding technics in severe tibial deformities of osteogenesis imperfecta

Tristan Langlais (Paris, France)

P65

What's in a Name? Prevalence of Metaphyseal Fractures in Children with Osteogenesis Imperfecta in the First Two Years of Life

Ella Riley (Sheffield, UK)

P66

Evidence for Metaphyseal Fractures Typical of Abuse in Osteogenesis Imperfecta: A Systematic Review

Ella Riley (Sheffield, UK)

P67

Bone Mass, Density, Geometry and Stress-Strain Index in Adults with Osteogenesis Imperfecta Type I, and their Associations with Physical Activity and Muscle Function Parameters

Marie Coussens (Ghent, Belgium)

P68

Typing with OI Type 5

Zoe Bowman Bayles (London, UK)

P69

Gene expression profiling of fetal mesenchymal stem cells during osteogenic differentiation

Emine Begum Gencer-Oncul (Stockholm, Sweden)

P70

Effect of Blood Flow Restriction Training on Bone, Muscle, Pain, and Fatigue in Adults with Osteogenesis Imperfecta type I: a Protocol Proposal of a Randomized Controlled Clinical Trial

Marie Coussens (Ghent, Belgium)

P71

Muscle Function Parameters in Adults with Osteogenesis Imperfecta type I: Preliminary Results

Marie Coussens (Ghent, Belgium)

P72

WNT1-related Osteogenesis imperfecta Type 15 and intramembranous calcification: subtle skull vault mineralisation defect only evident on 3D CT head.

Christine Burren (Bristol, UK)

P73

The Methodological Approaches Used to Understand the Psychosocial Experiences Of Parents of Children With Osteogenesis Imperfecta: A Review

Yu Chen Shi (Montreal, Canada)

P74

The Osteogenesis Imperfecta Variant Database: current state

Dimitra Micha (Amsterdam, Netherlands)

P75

Engaging the osteogenesis imperfecta (OI) community in patient centered outcomes research

Laura Tosi (Washington DC, USA)

P76

5-years experience of telescopic rods surgery for osteogenesis imperfecta patients in Russia
Alexey Rykunov (Moscow, Russia)

P77

The London Osteogenesis Imperfecta Team – Taking a Psychosocial Approach
Charlotte Barran (London, UK)

P78

A feasibility study exploring the suitability of the Bayley Scale of Infant and Toddler Development in assessing children from 16 days to 42 months with Osteogenesis Imperfecta who are under the care of the Metabolic Bone Team at Sheffield Children's NHS Foundation Trust
Carrie Marr (Sheffield, UK)

P79

Upper limb function in osteogenesis imperfecta: a scoping review
Jasmine Rocci (Oakville, Canada)

P80

Using virtual communication for rapid dissemination of COVID-19 information to patients with osteogenesis imperfecta
Laura Tosi (Washington, DC, USA)

P81

Quality of life and coping strategies in adults with OI
Ceú Barreiros (Lisbon, Portugal)

P82

Technique of telescopic nailing of the femur by retrograde approach (FRA) in patients with osteogenesis imperfecta: clinical and radiological results.
Zagorka Pejcin (Paris, France)

P83

Ability of Radiofrequency Echographic Multispectrometry in the assessment of bone mineral density in subjects with Osteogenesis Imperfecta
Antonella Al Refaie (Siena, Italy)

P84

The Severe Mouse Model of Osteogenesis Imperfecta Exhibits Compromised Cardiac Function
Brittany Lafaver (Columbia, USA)

P85

Gene expression signatures in bone and heart tissue of a Col1a1+/Mov13 mice identify it as a challenging animal model for mild Osteogenesis Imperfecta
Lidiia Zhytnik (Amsterdam, Netherlands & Tartu, Estonia)

P86

Vertebral fractures in Osteogenesis imperfecta with COL1A1 and COL1A2 -mutations
Bianca Link (Zurich, Switzerland)

P87

Patient osteoblast model for Osteogenesis Imperfecta using induced mesenchymal stem cells

Lauria Claeys (Amsterdam, Netherlands)

P88

Unfractured Care: Transforming Treatment Goals for the Infant with Severe Osteogenesis Imperfecta

Cristina McGreal (Wilmington, USA)

P89

Cardiovascular involvement in Osteogenesis Imperfecta - a Portuguese nation-wide cohort study (OI&Heart Study)

Ceu Barreiros (Sacavém, Portugal)

P90

Prevalence of tinnitus in adults with osteogenesis imperfecta

Temis Felix (Porto Alegre, Brazil)

P91

Landscape of Osteogenesis imperfecta in Brazil: data from the Brazilian Rare Disease Network

Temis Felix (Porto Alegre, Brazil)

P92

Using the Bayley Scales of Infant and Toddler Development to assess fine and gross motor skills in young children with severe Osteogenesis Imperfecta

Emilie Hupin (London, UK)

P93

Hearing in Osteogenesis imperfecta: a phenotype/genotype analysis

Temis Felix (Porto Alegre, Brazil)

P94

Defining Pathomechanisms and Unique and Common Molecular Signatures of MBTPS2 Osteogenesis Imperfecta

Pei Jin Lim (Zurich, Switzerland)

P95

Therapeutic Management of Olecranon Fractures in Paediatric Osteogenesis Imperfecta

Lisa Mills (Bristol, UK)

P96

Audiological follow up in Osteogenesis Imperfecta

Temis Felix (Porto Alegre, Brazil)

P97

Traumatic and non-traumatic cervical spine pathology in Severe, Complex and Atypical Osteogenesis Imperfecta

Catherine DeVile (London, UK)

P98

Proteomic analysis of the vertebral column in dominant OI zebrafish models to reveal biomarkers for phenotypic outcome

Andy Willaert (Gent, Belgium)

P99

Promoting Inclusion in Physical Education for Children with Osteogenesis Imperfecta: Preliminary Findings and Input Warranted for Guideline Creation

Galil Osman (Montreal, Canada)

P100

Osteogenesis Imperfecta Clinic Implementation of patient reported outcome measures (PROM): Implications, Pitfalls, Strategies, and Progress

Maureen Donohoe (Wilmington, USA)

P101

Evaluation of Radiolucent Lesions in Cortices of Long Bones in Osteogenesis Imperfecta Patients

Carolyn Cook (Chicago, USA)

P102

Design and Development of an E-Health Program for Youth with Osteogenesis Imperfecta – Teens OI

Raissa Passos dos Santos (Montreal, Canada)

P103

Skeletal Muscle Mitochondria Function in Osteogenesis Imperfecta Murine and G610C Mouse Models

Tara Crawford (Columbia, USA)

P104

Modification of the Osteogenesis Imperfecta Quality of Life Scale - Paediatric version (OIQoL-P) and Development of a Parent-Report version.

Claire Hill (London, UK)

P105

WHO-ICF Based Outcomes following Posterior Spinal Fusion with Instrumentation in a Child with OI Type III and Scoliosis- A Five-Year Follow-up

Frances Baratta-Ziska (New York, USA)

P106

Clinical and functional characterization of Osteogenesis Imperfecta Type XV: Single-centre Chinese Cohort

Michael To (Hong Kong, China)

P107

A retrospective review of 496 orthopedic surgeries in 223 Chinese patients of osteogenesis imperfecta

Michael To (Hong Kong, China)

P108

Clinical features and molecular characterization of Chinese patients with type XI osteogenesis imperfecta

Michael To (Hong Kong, China)

LM1

Self-perception on quality of life of children and adolescents with Osteogenesis Imperfecta attended in a University Hospital

Leilah Alves (Salvador-Ba, Brazil)

LM2

Type I collagen homotrimer exacerbates the impairment of bone structural properties caused by the osteogenesis imperfecta murine mutant allele, despite not itself causing bone fragility

Elizabeth Laird (Lowton, UK)

LM3

The Col1a2 +/G610C mutation causing osteogenesis imperfecta aggravate high-calorie diet-induced insulin resistance and bone deterioration in mice

Noam Levaot (Beer Sheva, Israel)

LM4

Osteogenesis imperfecta zebrafish models to test a new combined pharmacological approach

Antonella Forlino (Pavia, Italy)

LM5

Orbit: a randomized, double-blind, placebo-controlled, Phase 2/3 Study to assess the efficacy and safety of Setrusumab in pediatric and young adult participants with osteogenesis imperfecta

Michael Ominsky (Novato, USA)

LM6

Use of analgesics in patients with osteogenesis imperfecta in Denmark – a nationwide register-based cohort study

Lars Folkestad (Odense, Denmark)

LM7

Rigid intramedullary nailing of lower limb segments in adolescents with metabolic bone disease

Charlene Chin See (Sheffield, UK)

LM8

Chronic pain in adults with OI, its relationship with personality and the mediating role of assessment: a descriptive study.

Rubén Muñoz Cortés (Valencia, Spain)

LM9

Osteoporosis-pseudoglioma syndrome (OPPG) in two Bulgarian girls with c.2409_2503+79del mutation in LRP5 gene

Veselin Boyadzhiev (Varna, Bulgaria)

LM10

Microtensile properties of dry human bone extracellular matrix from individuals with Osteogenesis Imperfecta are not inferior to healthy controls

Michael Indermaur (Bern, Switzerland)

LM11

Natural history of fractures in children and adolescents with osteogenesis imperfecta type 1: A study from Western Australia

Kiranjit Joshi (Nedlands, Australia)

LM12

Results of telescopic nailing in children with osteogenesis imperfecta after minimum five-year follow-up

Ozan Ali Erdal (Istanbul, Turkey)

LM13

Effects of Losartan on Transforming-Growth-Factor- β and Angiotensin pathways in Osteogenesis Imperfecta

Mai Morita (Sheffield, UK)

LM14

Osteogenesis Imperfecta type VII: clinical variability and novel variants in CRTAP

André Travessa (Lisbon, Portugal)

LM15

Recurrent upper limb fractures: impact on quality of life and management challenges in two children with severe osteogenesis imperfecta

Emilie Hupin (London, UK)

LM16

Investigating mutations that cause osteogenesis imperfecta using molecular dynamics

Michael Jones (Liverpool, UK)