BALANCING LIFE WITH OI

Topical meeting on the impact of pain in osteogenesis imperfecta (OI)

9-10 June 2023 - Stockholm

PROGRAMME & ABSTRACTS
Organised by Osteogenesis Imperfecta Federation Europe (OIFE) in collaboration with the Swedish OI-organisation, SFOI
www.oife.org

This Networking event has received funding from the European Union’s Horizon 2020 research and innovation programme under the EJP RD COFUND-EJP N° 825575
Lack of understanding and poor management of pain severely impacts the quality of life of children and adults with osteogenesis imperfecta (OI).

Under the title “Balancing life with OI” we are organising a topical meeting on the neglected subject of the causes, assessment and management of pain in OI and its impact on physical and mental health, sleep, fatigue, mobility, relationships/families and work/life balance.

The conference programme includes research, case studies regarding the causes, assessment and management of pain in osteogenesis imperfecta and its impact on physical and mental health, sleep, fatigue, mobility, relationships/families and work/life balance. Comparisons with other rare bone diseases will also be made.

We look forward to your participation in the Conference and to spending some time with you in Stockholm!

*Lena Lande Wekre*
Senior Consultant and Conference Chair
TRS National Resource Center on Rare Disorders, Norway

*Ingunn Westerheim*
OIFE President
## Committees

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## Sponsors and supporters

We are very grateful to the following organisations for their support. Without them it would not be possible to organise this conference in this way.

**Grants, sponsorships and donations from industry**
- Alexion
- Angita Bio
- Azafaros
- Mereo Biopharma
- Quince Therapeutics
- UCB
- Ultragenyx

**Other Grants & Donations**
- European Joint Programme on Rare Diseases, Networking Support Scheme
- EURORDIS
- The OI Foundation

**Exhibitors**
- EuRR-Bone
- OICAN Wear
Credits - Opening Song “I've Got Pains”

The Shriners Hospital for Children in Montreal, Canada led and published a series of studies on the pain experienced by children and adults with OI.

At the time, graduate student Karl Williams, and the sibling of a former Shriners patient, wrote and recorded a song based on the research from the Shriners.

The song is called "I've got pains" and will be the opening song of the "Balancing Life with OI" conference, with permission from Shriners Hospital.

The animation video was done by Stephanie Smith.
Thursday 8 June
From 18:00  Arrival and registration

Friday 9 June
07:00 - 09:00  Breakfast and registration
09:00 - 10:30  Introduction to pain in OI and other rare bone conditions
   Moderators: Eva Åström (Sweden) / Ute Wallentin (Germany)

09:00  Welcome
   Eva Åström (Stockholm, Sweden) - Lena Lande Wekre (Oslo, Norway)
   Ingunn Westerheim (OIFE)

09:10  IS01 - Keynote lecture: What is pain?
   Audun Stubhaug (Oslo, Norway)

09:40  IS02 - What are the causes and different types of pain in OI?
   Jacqui Clinch (Bristol, UK)

10:00  IS03 - A multicenter study to evaluate pain characteristics in osteogenesis imperfecta: findings from the BBDC
   Mercedes Rodriguez Celin (Chicago, USA)

10:15  IS04 - Chronic pain in adults with OI and its relationship to appraisal, coping, and quality of life: A cross-sectional study
   Rubén Muñoz Cortés (Valencia, Spain)

10:30 - 11:00  Coffee break
11:00 - 13:00 **Assessment and measurement of acute and chronic pain**  
*Moderators: Jacqui Clinch (UK) / Inger-Margrethe Stavdal Paulsen (Norway)*

11:00 **Living with pain and OI – anonymous testimonies**  
*Read by Sara Innergård & André Wittwer*

11:10 IS05 - **How to assess: pros and cons of different tools**  
*Mercedes Rodriguez Celin (Chicago, USA)*

11:30 IS06 - **Neonatal pain**  
*Michael Bober (Wilmington, USA)*

11:45 IS07 - **The Assessment of Pain in Children with osteogenesis imperfecta**  
*Kelly Thorstad (Montreal, Canada)*

12:00 IS08 - **Assessing pain in adults with osteogenesis imperfecta**  
*Richard Keen (London, UK)*

12:15 - 13:00 **Oral Communications**  
*Moderators: Jacqui Clinch (UK) / Inger-Margrethe Stavdal Paulsen (Norway)*

12:15 S01 - **Sleep quality, excessive daytime sleepiness and increased risk for OSA in patients with osteogenesis imperfecta**  
*Linda Lušić (Split, Croatia)*

12:25 S02 - **Evaluation of mobility associated symptoms in Argentinian patients with Osteogenesis Imperfecta. Application of the STEMS tool (Screening Tool for Everyday Mobility and Symptoms for Skeletal Dysplasias)**  
*Mercedes Rodriguez Celin (Chicago, USA)*

12:35 S03 - **First results of using the Key4OI PROMIS questionnaires for measuring quality of life in children with OI, compared to children with other types of skeletal dysplasia**  
*Marjolein Verhoef (Utrecht, Netherlands)*

12:45 S04 - **The European Registry for Rare Bone and Mineral Conditions (EuRR-Bone): collecting patient reported outcomes on osteogenesis imperfecta**  
*Ana Priego (Leiden, Netherlands)*

12:50 S05 - **Pain and fractures in osteogenesis imperfecta: results from a survey of patients and caregivers**  
*Ali Skrinar (Novato, USA)*

12:55 S06 - **Sleep parameters in adults with OI and their associations with pain and fatigue**  
*Marie Coussens (Ghent, Belgium)*
13:00  Lunch and posters

Posters  (some oral communications may also be displayed as additional posters)

S18 - Learning through film: Contributing to a balanced life with OI  
Camilla LaHart (Oslo, Norway)

S19 - Chronic pain and fatigue in patients with multiple osteochondromas and Ollier disease: A systematic review  
Ariane Kwiet (Oslo, Norway)

S20 - Balancing function with pain in a child with Type V OI  
Frances Baratta Ziska (New York, USA)

S21 - The importance of nail follow-up: About a case  
Ana Maria Bueno Sanchez (Madrid, Spain)

S22 - Abstract withdrawn

S23 - Root resorption and dental maturity in children with osteogenesis imperfecta medicated with bisphosphonates  
Clara Sandibel, Garcete Delvalle (Madrid, Spain)

14:15 - 15.30 More than just bone pain

Moderators: Richard Keen (UK) / Lida Zhytnik (Estonia)

14:15 Living with pain and OI – anonymous testimonies  
Read by Anna Rossi & Willemijn Döpp - van Berkum

14:25 IS09 - Neuromuscular health and function in children with osteogenesis imperfecta  
Alex Ireland (Manchester, UK)

14:40 IS10 - Pain and its correlates with physical and psychological functioning in adults with OI  
Marie Coussens (Ghent, Belgium)

14:55 IS11 - Pain and basilar invagination  
Eva Åström (Stockholm, Sweden)

15:05 IS12 - Osteogenesis imperfecta and gastrointestinal pain – what do we (not) know?  
Lena Lande Wekre (Oslo, Norway)
15:15 - 15:30 **Oral Communications**  
*Moderators: Richard Keen (UK) / Lida Zhytnik (Estonia)*

15:15 S07 - Perceived dental care needs and concerns of individuals with osteogenesis imperfecta  
Coreen Kelday (Dundee, UK)

15:20 S08 - Biological plates in the surgical treatment of imperfect osteogenesis  
Ana Maria Bueno Sanchez (Madrid, Spain)

15:25 S09 - Intramedullary canal sclerosis - a possible complication of prolonged bisphosphonate therapy in children with osteogenesis imperfecta  
Željko Jeleč (Zagreb, Croatia)

15:30 - 16:00 Coffee break

16:00 - 16:30 **Debate: Can pain play a role as an endpoint and in evaluating new therapies?**  
*Moderator: Michael Bober (USA)*

Katarina Lindahl (Clinical Assessor, Swedish Medical Products Agency)  
Cathy Raggio (New York, USA)  
Ali Skirnar (Vice President Endpoint Development and Strategy, Ultragenyx)  
Inger-Margrethe Stavdal Paulsen (patient representative)

16:30 - 17:30 **How does pain in OI compare with other bone conditions?**  
*Moderators: Marelise Eekhoff (Netherlands) / Oliver Gardner (UK)*

16:30 IS13 - Pain and OI – similarities and differences between other bone conditions  
Alison Boyce (Bethesda, USA)

16:50 **Pain - examples from other rare bone conditions**

17:05 **Commonalities and differences - how can we work together on pain?**  
Marie Fahlberg (FOPSverige)  
Tenna Toft (XLH Alliance)  
Rebecca Tvedt Skarberg (OIFE)  
Meryl Chambers (Soft Bones HPP UK Foundation)  
Liana la Forgia (Italian Patient Association on Multiple Osteochondromas)

17:30 - 18:00 **Oral Communications**  
*Moderators: Marelise Eekhoff (Netherlands) / Oliver Gardner (UK)*

17:30 S10 - Pain and Quality of Life in patients with Fibrous Dysplasia/McCune Albright Syndrome: a prospective follow up study  
Oana Bulaicon (Leiden, Netherlands)
17:40 S11 - Working life with rare diagnosis
Brede Dammann (Oslo, Norway)

17:50 S12 - Pain rehabilitation of patients with a rare skeletal dysplasia – adapting to their special needs within existing rehabilitation programs
Ariane Kwiet (Oslo, Norway)

17:55 S13 - Use of medical care by individuals with osteogenesis imperfecta in the Netherlands for neurological and pain-related issues
Silvia Storoni (Amsterdam, Netherlands)

18:00 Conference ends

19:15 - 20:00 Welcome reception (for all)

20:00 - 22:00 Conference dinner (tickets required)

Saturday 10 June

07:00 - 09:00 Breakfast

08:00-08:45 Morning Session: Update on Clinical Trials in OI
Moderator: Alex Ireland (UK) / Taco van Welzenis (Netherlands)

Project Saturn – a Real-World Evidence and data collaboration with existing European datasets in OI to support future therapies
James Clancy (Mereo BioPharma)

Cosmic & Orbit: an update on Ultragenyx trials in OI
Ali Skirnar (Ultragenyx)

BOOST Pharma: Stem cells as a treatment for OI, the BOOSTB4 clinical trial and beyond
Lilian Walther Jallow (Boost Pharma)

TGF-β antibody for the treatment of osteogenesis imperfecta
Cemre Robinson (Sanofi)

09:00-11:00 The IMPACT of pain in OI
Moderators: Cathy Raggio (USA) / André Wittwer (Norway)

9:00 Living with pain and OI – anonymous testimonies
Read by Lars Nesset Romundstad & Stephanie Claeyes
9:10 IS14 - The IMPACT Survey: an international collaborative research initiative for the OI community
*Samantha Prince (Abingdon, UK)*

9:25 IS15 - Self-reported prevalence and impact of pain in persons with osteogenesis imperfecta (OI): some perspectives from the IMPACT study
*Lena Lande Wekre (Oslo, Norway)*

9:40 IS16 - Understanding the connection between pain in OI and mental health, and the impact on sleep and fatigue
*Andrew Wiese (Houston, USA)*

10:00 **Balancing ambitions with pain in OI**
*Karen Braitmayer (USA) / Jacob Wittorff (Denmark)*

10:15 IS17 - Parenting a child in pain
*Kis Holm Laursen (Denmark)*

10:30 IS18 - Pain’s impact on relationships and sexuality
*AnnBett Kirkebæk (Aarhus, Denmark)*

10:45 IS19 - The Pain and OI Survey
*Michael Stewart (OIF, USA)*

11:00 - 11:20 **Coffee break**

11:20-11:50 **Basic science and pain**
*Moderators: Dimitra Micha (Netherlands) / Taco van Welzenis (Netherlands)*

11:20 IS20 - Pain mechanisms in heritable collagen disorders: lessons learned from the Ehlers-Danlos Syndromes
*Fransiska Malfait (Ghent, Belgium)*

11:50-12:50 **Managing pain in OI (part 1)**
*Moderators: Kis Holm Laursen (Denmark) / Lena Lande Wekre (Norway)*

11:50 IS21 - How we work in the MDT pain clinic: Bristol and Oslo
*Jacqui Clinch (Bristol, UK) / Audun Stubhaug (Oslo, Norway)*

12:10 IS22 - Modalities to deal with pain caused by fractures and surgeries
*Cathy Raggio (New York, USA)*

12:30 IS23 - Pharmacological pain management
*Richard Keen (London, UK)*
12:50 - 14:00 Lunch and posters (see Friday for list)

14:00-15:45 Managing pain in OI (part 2)
  Moderators: Mercedes Rodriguez Celin (USA) / Rebecca Tvedt Skarberg (Norway)
  
  14:00 IS24 - Therapeutic pain management principles in OI
  Sophie Barlow (London, UK)
  
  14:20 IS25 - Physical activity in OI: A good natural medicine for pain and health in OI
  Miguel Rodrigues Molina (Madrid, Spain)
  
  14:35 IS26 - Creation of educational resources for children with osteogenesis imperfecta experiencing acute and chronic pain
  Kelly Thorstad (Montreal, Canada)
  
  14:50 IS27 - A toolbox for pain in OI: empowering patients to take charge of their health
  Ariane Kwiet (Oslo, Norway)

15:05 - 14:45 Oral Communications
  Moderators: Mercedes Rodriguez Celin (USA) / Rebecca Tvedt Skarberg (Norway)
  
  15:05 S14 - Transition and Follow-up in Adult OI
  Jannie Hald (Aarhus, Denmark)
  
  15:15 S15 - Pain and its biopsychosocial impact in children with skeletal dysplasias
  Saunya Dover (Ottawa, Canada)
  
  15:25 S16 - 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)
  
  15:35 S17 - Impact of advanced therapy on pain in OI
  Clara Rodriguez (Bilbao, Spain)

15:45 - 16:00 Closing remarks and pink elephants!
  Ingunn Westerheim (OIFE, Norway)
  Moderator: Lena Lande Wekre (Norway)

16:00 - 16:30 Coffee break and departure for those not attending OIFE and SFOI meetings
**FOR MEMBERS OF OIFE, SFOI & INVITED GUESTS ONLY**
*(separate registration required)*

16:30-19:00  **OIFE Annual General Meeting**
Swedish Association for Osteogenesis Imperfecta (SFOI)

20:00-22:00  **3 course dinner** (included) OIFE & SFOI

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**Sunday 11 June**

**FOR MEMBERS OF OIFE, SFOI & INVITED GUESTS ONLY**

10:00-13:00  **Workshops** (OIFE AGM continues if needed)

13:00  **Grab and go lunch**
DISCLAIMER

Abstracts are published as submitted by the authors. All parties will make reasonable efforts to present the educational subject matter in a scientific, balanced and unbiased way.

Statements and opinions given are informational only and are not made or given as a warranty.

The views, opinions and statements made at the conference are solely those of the presenters and may not reflect the views of the Organisers. Furthermore, participants should bear in mind that presenters may have vested interests in the concepts and products they discuss.

ABBREVIATIONS

Invited speakers
Abstracts from invited speakers are listed as IS + number.

Speakers
Abstracts based on abstract submissions are listed as S + number.
Pain is defined as "An unpleasant sensory and emotional experience associated with, or resembling that associated with, actual or potential tissue damage." (IASP). Pain is always a personal experience that should be respected, and pain is influenced to varying degrees by biological, psychological, and social factors. All individuals learn through their life experiences the concept of pain. Most often, pain serves an adaptive role. However, chronic pain may have adverse effects on function and social and psychological well-being. There is a huge difference in pain sensitivity between subjects. At the same time, pain sensitivity may change rapidly within the same individual, e.g. after repeated acute injuries. Most subjects have lowered pain thresholds after a single injury. After repeated pain incidents, secondary problems affecting function, sleep and quality of life are common. When pain becomes chronic, it loses the functional adaptive role, and becomes a negative part of life, often heavily affecting function and quality of life.

Pain may be classified according to the mechanism involved as either nociceptive, neuropathic or nociplastic. Nociceptive pain is the typical well understood pain caused by tissue damage, like a fracture. Neuropathic pain is caused by damage to the neural system, like a spinal cord injury or a nerve injury. Nociplastic pain describes the changed central nervous system's interpretation of normal peripheral nervous input. Nociplastic pain is a recent concept that is slightly controversial.

The pain diagnosis and the pain classification is crucial in order to find the best treatment. Nociceptive pain resembles most acute pains and can be relieved by anti-inflammatory drugs like paracetamol, NSAID and corticosteroids. In addition, short time use of opioids may be very effective. Treatments for neuropathic and nociplastic pain will also be reviewed briefly.

When pain becomes chronic, the aim changes from pain reduction into increased function and quality of life. That means that drugs have a more limited place while other lifestyle changes are potentially more important.

Non-pharmacological treatment is the most important approach in chronic pain. Possible treatments will be reviewed.
OI-individuals may have all types of pain mentioned above. A correct pain analysis and pain classification is needed to guide the individual and the health professional to the best possible treatment.

**Disclosure**
None declared

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**IS02**

*What are the causes and different types of pain in OI?*

**Jacqui Clinch**
*Bristol Royal Hospital for Children, Bristol, United Kingdom*

Rare bone diseases can cause different types of pain across the lifespan. This talk covers the scientific background of musculoskeletal pain in all types of osteogenesis imperfecta and explores the relationships between pain and physical & psychological wellbeing.

**Disclosure**
None declared

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**IS03**

*A multicenter study to evaluate pain characteristics in osteogenesis imperfecta: findings from the BBDC*

**Mercedes Rodriguez Celin**¹², Karen Kruger³⁴, Angela Caudill³, Chaya Murali⁵, Sandesh Nagamani⁵, Members of the Brittle Bone Disorders Consortium BBDC⁵, Peter Smith³, Gerald Harris⁴
¹ Shriners Hospital for Children, Chicago, IL, USA. ² University of Illinois, Chicago, IL, USA. ³ Shriners Hospital for Children, Chicago, IL, USA. ⁴ Marquette University, Milwaukee, WI, USA. ⁵ Baylor College of Medicine, Houston, TX, USA

**Objective**
The objective was to describe pain characteristics and treatments used in individuals with varying severity of osteogenesis imperfecta (OI) and to investigate pain-associated variables.
**Methods**

This work was derived from a multicenter, longitudinal, observational, natural history study of OI conducted at 12 clinical sites of the NIH Rare Diseases Clinical Research Network’s Brittle Bone Disorders Consortium. Children and adults with a clinical, biochemical, or molecular diagnosis of OI were enrolled in the study. We did a cross-sectional analysis of chronic pain prevalence, characteristics, and treatments used for pain relief and a longitudinal analysis to find the predictors of chronic pain.

**Results**

We included 861 individuals with OI; in 41.8%, chronic pain was present with similar frequency across OI types. Back pain was the most frequent location, followed by multiple bones and joints.

Nonsteroidal anti-inflammatory drugs, followed by bisphosphonates, were the most common treatment used. Individuals with OI combined pharmacological and non-pharmacological therapies for pain relief. Participants with chronic pain missed more days from school or work/year and performed worse in all mobility metrics than participants without chronic pain. The variables more significantly associated with chronic pain were age, sex, positive history of rodling surgery, scoliosis, other medical problems, assistive devices, lower standardized height, and higher body mass index. The predictors of chronic pain for all OI types were age, use of a wheelchair, and the number of fractures/year.

**Conclusions**

Chronic pain is prevalent in OI across all OI types, affects mobility, and interferes with participation. Multiple covariates were associated with chronic pain.

**Disclosure**

No conflict of interest

**IS04**

*Chronic pain in adults with OI and its relationship to appraisal, coping, and quality of life: A cross-sectional study*

Rubén Muñoz Cortés ¹, José Francisco Soriano Pastor ², Vicente Monsalve Dolz ³

¹Fundación AHUCE, Valencia, Spain. ²Universidad de Valencia, Valencia, Spain. ³Hospital General Universitario, Valencia, Spain
Chronic pain is a common experience in osteogenesis imperfecta (OI). However, there are few studies on this topic in the field of psychology as a discipline. The aim of this study is to describe the frequency and characteristics of chronic pain in a large sample of adults with OI, as well as its relationship with clinical, sociodemographic, psychological, and quality of life variables.

A cross-sectional study was conducted in a sample of 418 adults with OI who responded to an online questionnaire battery. Sociodemographic and clinical variables, pain parameters, personality characteristics, pain appraisal, coping strategies, interference with daily activities, and health-related quality of life were evaluated. A descriptive, correlational, and mean contrast analysis was performed.

Up to 83% of the sample reported experiencing pain frequently. Both pain frequency and intensity were related to the accumulation of fractures over the years \( (P < 0.05) \), but were independent of other variables such as the severity of the pathology or the use of bisphosphonates. Participants with higher levels of neuroticism experienced pain more frequently \( (P < 0.000) \) and with greater intensity \( (P < 0.000) \), while higher scores of extroversion were related to a decrease in pain frequency \( (P < 0.000) \) and intensity \( (P < 0.000) \). A higher threat appraisal of pain was associated with an increase in perceived pain intensity and interference with daily activities, as well as a decrease in physical and mental health \( (P < 0.001) \).

Chronic pain is a frequent condition in adults with OI, regardless of the severity of the pathology. It interferes with their usual activities and has an impact on their quality of life. Personality traits, as well as how participants appraise their pain, also have an influence on its intensity and consequences. Interventions aimed at training in strategies to manage pain appraisals could potentially improve adaptation to chronic pain.

**Disclosure**
None declared.

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**IS05**

**How to assess pain: pros and cons of different tools**

**Mercedes Rodriguez Celin**

Shriners Hospital for Children, Chicago, USA. University of Illinois, Chicago, USA
Objective
Pain is a subjective unpleasant experience that can be disabling, affecting the quality of life, participation, function, and physical and psychosocial well-being. The growing need for an improved understanding of pain requires a better assessment and evaluation. Pain is understudied in OI, and no standard assessment method has been described.

Method
This invited talk will provide a brief literature overview of the different types of pain and causes of pain in OI. Finally, we will review the different methods to assess pain in children and adults.

Results
Understanding the pros and cons of using different assessment methods could be helpful in the decision to improve pain assessment in OI. Following the literature recommendations, we will encourage the use of validated, age-appropriate, and, if possible, multidimensional tools for pain assessment appropriate to different settings.

Conclusion
A better assessment of pain is the first step to improving the clinical management of pain in OI.

Disclosure
No conflict of interest

IS06
Neonatal pain

Michael Bober, Ricki Carroll
Nemours Children’s Health, Wilmington, USA

For neonates with Osteogenesis Imperfecta, (OI) pain management is critical. Through experience at our center, we have developed a standardized approach that has been successful in optimizing survival for infants with moderate and severe OI. Delivery of these infants is often complicated by the occurrence of fractures. The likelihood of fractures is affected by the severity of the infant’s OI, with at least 90% of newborns with Type III OI and 50% of newborns with Type IV OI experiencing at least one fracture directly related to delivery.
Fractures not only cause discomfort but also can have a direct impact on an infant’s respiratory status and overall clinical picture. Therefore, adequate pain control is imperative to both survival and well-being of infants with OI.

An opioid infusion is generally required for adequate pain control during the first few days to weeks of life, depending on the number of and type of fractures present. This is especially important if rib fractures are present, not only for comfort but also to decrease respiratory splinting and promote respiratory efficiency. Additional as-needed doses of opioids should be used whenever pain scores are elevated.

At approximately 7-10 days of life, we expect to see woven bone formation at the site of birth fractures develop into a visible callous and stabilize the fracture site. Around this time, infants will typically begin moving their extremities more and showing signs of recovery. Once there is evidence of fracture healing clinically, weaning the opioid infusion can begin, with the ultimate goal of discontinuation. Once acute fracture pain has dissipated, management of more generalized bone pain is important; therefore, we advocate starting the first pamidronate cycle as soon as is practical. Fracture mitigation should pervade all aspects of care, beginning at birth.

Strategies include: signs, egg crate mattresses, loose-fitting clothing, avoidance of sleeves, strict adherence to clustering hands-on care, including position changes and diaper changes; and avoiding unnecessary movement, such as daily weights. Despite careful handling and optimized medical care, fractures may still occur. The management of these fractures is positioning and immobilization with a lightweight soft wrap for the extremity.

Disclosure
None declared

IS07

The Assessment of Pain in Children with Osteogenesis Imperfecta

Kelly Thorstad

Children with OI will experience acute and chronic pain, which may interfere with their daily functioning and quality of life. Frequent fractures are one of the causes of acute pain in children. More debilitating may be chronic pain due to accumulated injuries over time and bone deformities, which interferes with sleep, mobility, and participation in school and other activities.
Pain being a complex, multidimensional, and subjective phenomenon, there is a need for tools to assess pain adequately and comprehensively in children with OI. Hence, our team at the Shriners Hospitals for Children®-Canada led literature reviews, and conducted pain studies to understand the pain experiences of children, adolescents, and adults with OI and determine how pain was being assessed. Our research led to the revelation of the paucity of standardized methods to assess pain in OI. In this talk, we share ways of assessing pain in children with OI in practice.

Disclosure
None declared

IS08
Assessing pain in adults with osteogenesis imperfecta

Richard Keen
Royal National Orthopaedic Hospital, Stanmore, United Kingdom

Pain is a common symptom in adults with osteogenesis imperfecta (OI). Acute pain will be seen in patients experiencing a fracture, and this pain will often improve with appropriate orthopaedic management. Chronic pain is, however, more complex and can be due to a number of factors. Deformities after fractures to the appendicular skeleton and spine can result in impaired muscle function and fatigue.

There may also be the development of premature degenerative joint disease, with associated pain and stiffness. Pain may also arise from ligaments and soft tissue, especially in those with features of hypermobility. Fatigue and poor sleep pattern, which are a common features in patients with chronic musculoskeletal conditions, can negatively impact on a patient’s perception of pain.

In contrast to research in the paediatric population, there are a relatively small number of studies that have examined pain in adults with OI. These studies do find that OI patients report significantly higher rates of pain when compared to the general population and that these symptoms impact on their daily life, mobility, participation, work and social relationships. The studies have generally utilised a number of quality of life questionnaires and pain specific tools, including EQ-5D, SF-36, BPI and Visual Analogue Scales.

At present there is no assessment tool in general use that is specific for people with OI.
The challenge in the clinic is finding a tool that can be used in a time-efficient manner and can help both the patient and clinician characterise pain which may be amenable to treatment or lifestyle interventions. Virtual or on-line applications that patients can complete remotely may also be better as they can give an overview of pain symptoms over time.

**Disclosure**
None declared

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**IS09**

**Neuromuscular health and function in children with osteogenesis imperfecta**

**Alex Ireland**  
*Manchester Metropolitan University, Manchester, United Kingdom*

The type I collagen affected by osteogenesis imperfecta (OI) is also found in muscle and tendon. The impaired quality and/or quantity of this collagen in children with OI affects multiple components of neuromuscular function, including balance, strength, and fatigue. This has consequences for skeletal development and risk of falls and fractures, as well as aspects of daily living such as mobility and even educational attainment. The intrinsic biomechanical and physiological factors that underpin these deficits may include altered muscle mechanics, and altered calcium handling. Consequences of OI such as bone and joint pain and altered cardiovascular and respiratory function likely also contribute, although this remains unexplored. Similarly, little is known about relevant environmental exposures such as physical inactivity or altered nutrition. A small number of studies have investigated the effects of physical therapy and other interventions to improve function, but current evidence is limited. This talk will describe our current understanding of neuromuscular health and function in children with OI, its causes, consequences and potential treatments. It will also identify key areas where the current evidence base is weak, and where more detailed studies could provide important insights.

**Disclosure**
None declared
IS10

Pain and its correlates with physical and psychological functioning in adults with OI

Marie Coussens¹, Patrick Calder³, Inge De Wandele², Fransiska Malfait²³, Tash Pocovi⁴, Verity Pacey⁴
¹Department of Rehabilitation Sciences and Physiotherapy, Ghent University, Ghent, Belgium. ²Centre for Medical Genetics, Ghent University Hospital, Ghent, Belgium. ³Reference Centre for Rare Bone, Calcium and Phosphate Disorders, Ghent University Hospital, Ghent, Belgium. ⁴Department of Health Professions, Macquarie University, Sydney, Australia

Objectives
To evaluate pain levels in adults with osteogenesis imperfecta (OI) and to identify their correlates with physical and psychological parameters.

Methods
Sixty-eight adults with OI (45 type I, 12 type III, 10 type IV, 1 type V; 60% females), mean age 42 years (±13.9), were recruited in Belgium and Australia. Pain severity (visual analogue scale (VAS)) and painful body surface (Margolis pain diagram) were assessed before performing physical (30 seconds chair rise (30s CRT), 6 minute walking, and hand grip strength test) and joint hypermobility tests. Furthermore, pain severity (multidimensional pain inventory (MPI)) and painful body surface were kept in a diary during seven days together with anxiety and depression (Hospital Anxiety and Depression Questionnaire) and fatigue (Checklist Individual Strength) levels. Health-related quality of life (HR-QoL; Arthritis Impact Measurement Scale 2) and Health Status (36-Item Short Form Survey) were also assessed. Data are shown as median [IQR]. Association (Spearman correlation) and non-parametric analyses were performed.

Results
Median painful body surface was 4.25% [2.25; 6.50], pain severity 2.00 [2.00; 3.00] out of 10 (VAS) and 2.72 [1.86; 3.71] out of 18 (MPI). Painful body surface was negatively associated with 30s CRT (rs=-0.438; p<0.001), health status (role limitations due to physical health and emotional problems, and general health; rs=-0.512 to -0.321, p≤0.034) and HR-QoL (rs=-0.432, p=0.003), and positively associated with depression (rs=0.303, p=0.043) and fatigue levels (rs=0.362, p=0.016). Pain severity was negatively associated with HR-QoL (rs=0.809, p<0.001), health status (physical functioning, role limitations due to physical health, energy/fatigue, social functioning, and general health; rs=-0.535 to -0.349, p≤0.022), and positively associated with anxiety (rs=0.533, p<0.001), depression (rs=0.574, p<0.001) and fatigue levels (rs=0.627, p<0.001).
Non-parametric tests showed no significantly different pain levels or painful body surface between sexes, OI types or individuals with or without generalized joint hypermobility.

**Conclusion**
Greater severity and higher extent of pain in adults with OI is associated with lower health-related quality of life and health status, and higher fatigue and depression levels. Additionally, painful body surface is negatively associated with lower limb strength. Hypermobility, sex, and severity of OI probably play no significant role in their pain.

**Disclosure**
None declared.

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**IS11**

**Pain and basilar invagination**

**Eva Åström**

Karolinska University Hospital, Stockholm, Sweden. Karolinska Institutet, Stockholm, Sweden

Skull base anomalies in OI can be present at birth or develop later in life. Most individuals with OI will never have skull base problems. Basilar invagination is a development anomaly with migration of the upper cervical spine and odontoid peg into the base of the skull and penetrating the foramen magnum. In the general population estimated prevalence 1%. It is more common in individuals with softer bone and is correlated to the severity of OI (to relative or absolute macrocephaly, to short stature, to OI types 3, 4 and 5 and to DGI). BI can lead to brain stem and cerebellum compression, disturbed CSF flow and cerebral blood flow and affect cranial nerves. Basilar impression is an upward infolding of the foramen magnum area into the skull (sometimes referred as “the acquired form of Basilar invagination”. Platybasia is a flattening of the skull base with a basal angle >145 degrees. They can exist together or in isolation and platybasia is most common. Diagnosis is often made with routine lateral skull X-ray and in case of typical symptoms MR is important.

BI is often asymptomatic and generally progress slowly in childhood. It does not always progress but the incidence increases with age. Neurologic signs may be present before symptoms (nystagmus, facial spasms, cranial nerve paresis, proprioceptive deficits). Pain usually occurs due to incipient ventral brainstem compression where the most common symptom is occipital headache (initially only at coughing or sneezing or with movement) or neck pain.
Other symptoms are vertical nystagmus, dysphagia, change in facial sensation (numbness), upper extremities weakness, downbeat gaze, central sleep apnoea, torticollis, ataxia...). Untreated severe BI can lead to secondary hydrocephalus and brain stem compression with respiratory problems and sudden death.

Prevention
Supine position of infants with OI until motor stability is achieved? Early physiotherapy? Early treatment with bisphosphonates?

Therapy
Non-addictive analgesics, relieve the compression by operation (ventral decompression and posterior fixation), (or posterior atlantoaxial distraction and fixation).

Disclosure
None declared

IS12

Osteogenesis imperfecta and gastrointestinal pain – what do we (not) know?

Lena Lande Wekre

TRS National Resource Center for Rare Disorders, Sunnaas Rehabilitation Hospital, Norway

Background
Patient-reported concerns, and clinical experience, indicate that gastrointestinal (GI) problems are quite common in the OI population. However, quantitative information regarding prevalence and severity of the GI manifestations is limited.

Objective of this talk
To give a short overview about what we know, and what we do not know, about GI manifestations and pain in OI.

Results
Stomach problems are described as common in OI. These include gastric acid reflux, chronic constipation and recurrent abdominal pain, which are all relatively frequent, reported. GI pain is also reported as more frequent in those who have acetabular protrusion than in those who do not have protrusion.
Conclusion
To give the best treatment, increased knowledge about the type of gastric manifestations, the aetiology of the problems, degree and type of pain, and whether there are any age differences, is needed.

Disclosure
None declared.

IS13

Pain and OI – similarities and differences between other bone conditions

Alison Boyce
National Institutes of Health, Bethesda, USA

Pain is common in patients with rare bone disorders. Although these disorders have many different causes, they share similarities related to pain and its treatment. Therefore, research into the mechanisms and management of pain in one disorder may improve our ability to manage a broad range of patients. In this presentation, we will discuss causes of pain in rare bone disorders other than OI.

We will compare and contrast pain in OI with these disorders, and discuss how this informs our understanding of bone biology, research priorities, and treatment strategies.

Disclosure
NIDCR receives research funding from Amgen, Ultragenyx, and Kyowa Kirin
The IMPACT Survey: an international collaborative research initiative for the OI community

Samantha Prince¹, Lena Lande Wekre², Michael B Bober³, Cathleen Raggio⁴, Oliver Semler⁵, Ingunn Westerheim⁶, Tracy Hart⁷, Taco van Welzenis⁶, Maria Rapoport¹, Frank Rauch⁸
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Objectives
The IMPACT Survey was designed to address knowledge gaps about life with OI and capture a diverse range of community perspectives across age groups, geographies, and backgrounds. The questionnaire explored the clinical, humanistic, and economic impact of OI to support future advocacy work, policy initiatives, and better healthcare.

Methods
An international Steering Committee of clinical experts, OI community– and industry representatives was set up to guide survey development and advise on the most relevant questions for the OI community whilst maintaining scientific rigour. Existing knowledge gaps were identified by comprehensive review of the published OI literature before developing the survey to address some of those gaps. The survey design was shaped and validated by clinical experts and OI community members from around the world and translated into 8 languages. Online fielding was conducted from July–September 2021 and recruitment efforts were driven by OIFE and OIF networks through a variety of channels.

Results
2,208 self-reported and 780 proxy responses were captured across 68 countries. Populations include 1,575 adults (≥18 years) with OI, 263 adolescents with OI (12–17 years), 474 children with OI (<12 years), 710 caregivers (with and without OI themselves), and 116 close relatives of people with OI.

The data is owned by a management committee comprised of representatives from the OIFE, the OIF and academia. Data analysis is ongoing and core results will be published and shared with the OI and research communities over the coming year.

Conclusion
This dataset represents the largest of its kind to date, includes diverse respondent profiles and a range of topics not previously explored. The scope of this collaborative research
effort enables data analysis and application for the benefit of the community for years to come, opening the door for new opportunities to deepen our understanding of the true impact of living with OI.

Disclosure
SP and MR are employees of Wickenstones Ltd, Abingdon, United Kingdom. MB has received grants from Ultragenyx Pharmaceuticals Inc, Novato, USA, and the Osteogenesis Imperfecta Foundation. TH serves as the Chief Executive Officer of the Osteogenesis Imperfecta Foundation and has received unrestricted educational grants from Mereo BioPharma Group London, United Kingdom and Ultragenyx Pharmaceuticals Inc, Novato, USA. OS has participated in a national advisory board for Mereo BioPharma Group London, United Kingdom. FR has received study contracts for experimental preclinical studies with Precithera Inc, Quebec, Canada, Mesentech Inc, Vancouver, Canada and Catabasis Pharmaceuticals Inc, Cambridge, USA. He has participated in advisory boards for Ultragenyx Pharmaceuticals Inc, Novato, USA, Sanofi S.A. Paris, France, Novartis International AG, Basel, Switzerland and Mereo BioPharma Group, London, United Kingdom. FR has received a speaker fee from Ultragenyx Pharmaceuticals Inc, Novato, USA for a lecture and received a donation of experimental drugs for a preclinical study from Acceleron Pharma Inc, Cambridge, USA. CR received an institutional grant from BioMarin Pharmaceuticals Inc, Novato, California, has participated in advisory boards for Ultragenyx Pharmaceuticals Inc, Novato, USA and sits on the medical board of the Osteogenesis Imperfecta Foundation. TW and IW hold leadership positions in the Osteogenesis Imperfecta Foundation Europe, which has received grants from Mereo BioPharma Group, London, United Kingdom.

IS15

Self-reported prevalence and impact of pain in persons with osteogenesis imperfecta (OI): some perspectives from the IMPACT study

Lena Lande Wekre, Samantha Prince, Cathleen Raggio, Michael B Bober, Oliver Semler, Ingunn Westerheim, Tracy Hart, Taco van Welzenis, Maria Rapoport and Frank Rauch

Objectives
The IMPACT survey aimed to collect the most comprehensive self-reported dataset on the quality of life and experiences of persons, all ages, with OI. A better understanding of the real impact of OI on affected individuals, including quality of life and clinical sequelae, is instrumental to improving OI care and develop relevant guidelines.
Methods
Together with the OI Federation Europe (OIFE) and OI Foundation (OIF) we have developed an international survey. It was fielded using an online platform and aimed at adults (aged ≥18 years) or young people (aged ≥12–17 years) with OI, caregivers (with or without OI) and other close relatives of children or adults with OI. Questions covered the patient healthcare journey, worries and concerns, and impact on individuals, their families, and finances. Data were cleaned, coded, and analysed using StataSE 17.0.

Results
The most common conditions experienced by the adult cohort with OI (1,440 adults - 70% female, mean age 43.3 years) were pain (81.8%), fatigue (66.9%) and soft tissue problems incl. muscles, tendons and ligaments (54.8%). Of all conditions experienced in the past 12 months, mental health issues was most commonly ranked as moderately or severely impactful (by 77.7% of those experiencing the condition), followed by reproductive health issues (76.3%), sleep disturbance (72.7%) and soft tissue problems (71.5%).

The most common self-reported musculoskeletal characteristics in an adolescent cohort (92 adolescents – 55% females, mean age 14.8 years) were pain (81.5%), fatigue (65.2%) and scoliosis or other bone problems (59.8%)

Conclusion
This presentation will give an overview of the findings regarding pain, and discuss the challenges and opportunities from different perspectives.

Disclosure
None declared

IS16
Understanding the connection between pain in OI and mental health, and the impact on sleep and fatigue

Andrew Wiese, Hannah Cho, Whitney Shepherd
Baylor College of Medicine, Houston, USA

Research and patient care for osteogenesis imperfecta (OI) has historically focused on disease presentation and management of physical health symptoms. Relative to other health populations, assessment, characterization, and interventions designed for OI-specific psychosocial concerns remain understudied and poorly understood.
This has resulted in providers being ill equipped to identify and address areas of concern specific to OI outside of physical health, including symptoms of anxiety, depression, sleep, and psychological impacts associated with injury and pain. In this presentation, Dr. Andrew Wiese will discuss existing health psychology literature related to OI, along with ongoing research being conducted in the United States at Baylor College of Medicine and collaborating sites involved in the Brittle Bone Disorder Consortium (BBDC).

As a part of a multi-phased study, he will review preliminary findings from Phase I qualitative interviews with various OI stakeholder groups, including adult and paediatric individuals with OI, caregivers, and healthcare providers. He will additionally provide Phase II updates on longitudinal assessments of various psychological constructs being collected as part of the BBDC’s Natural History Study.

In reviewing Phase III, he will discuss efforts underway to assess relationships between various psychological predictors of adverse health outcomes, as well as factors that may buffer against adverse outcomes among those diagnosed with OI.

Finally, the importance of developing psychological interventions for those with OI and other health conditions will be discussed as a future priority for OI researchers and clinicians.

Disclosure
None declared

IS17

Parenting a child in pain

Kis Holm Laursen
Det Unikke Barn, Aarhus, Denmark

The pain in a child with OI has an impact on the entire family. The parent might be in despair because they cannot take the pain away or stop the fracture. Siblings live in the fear of causing fractures/pain or being an extra burden for the parents.

Children in pain will make the parents alert and the alertness in parents and the child will affect the child’s pain experience.

Siblings will be affected by seeing their sister or brother in pain and the parents in fear and alertness. Siblings can have a hard time expressing their needs for normal activities. Siblings can hold back feelings or thoughts to protect their parents.
Parents need inspiration and new skills to create a family with love and understanding for the entire family’s needs.

Disclosure
None declared

IS18

Pain’s impact on relationships and sexuality

AnnBett Kirkebaek
FitnessX ApS, Aarhus, Denmark

Men and women with (chronic) pain may find it difficult to perceive themselves as sexually attractive, when the pain is unbearable and requires every effort in daily life.

I was very interested in investigating how pain (due to OI) in one party affects the relationship, both emotionally and sexually, and what can be done to alleviate the pain.

I found that physical exercise can provide pain relief for those with OI. And that doing sports can positively affect your body’s health in many ways, as for example creating physical awareness and muscle tension.

The talk will introduce the training method; Pilates and give examples on how Pilates can strengthen the body, alleviate the pain, and prepare the body for sexual intimacy.

I also found that the conversation about the pain, between the parties in the relationship, is crucial and sometimes even a better pain relief overall!

The feeling of certainty/security and understanding are the most decisive factors in a relationship to be able to preserve and perhaps also rebuild the emotional closeness.

The talk will introduce examples on what might strengthen certainty/security and understanding in the sexual relationship and examples that might weaken it. And the talk will do this based on the main topic of pain.

Disclosure
None declared
**IS19**

**The Pain and OI Survey**

**Michael Stewart**  
*Osteogenesis Imperfecta Foundation (OIF)*

In January 2023, the Pain and OI Project, a group co-led by the Osteogenesis Imperfecta Foundation (OIF) and the Osteogenesis Imperfecta Federation Europe (OIFE), created and administered a survey where any adult with OI across the globe could share information about their experience of pain and how it impacts their lives. In a few months, the survey collected 1086 responses from individuals in 49 countries. Michael Stewart, OIF Director of Education, will discuss its creation and present some initial results. The survey results will be further analyzed by a research team at Baylor College of Medicine in Houston, Texas.

**Disclosure**  
None declared

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**IS20**

**Pain mechanisms in heritable collagen disorders: lessons learned from the Ehlers-Danlos Syndromes**

**Fransiska Malfait**  
*Center for Medical Genetics, Ghent University Hospital, Ghent, Belgium*

Heritable disorders of connective tissue (HDCT) are a heterogeneous group of conditions caused by genetic defects in the structure and synthesis of extracellular matrix (ECM) molecules such as collagen, elastin, or proteoglycans. Chronic, intractable pain is a frequent complaint of patients with HCTD, with significant overlap regarding pain mechanisms, phenotypes, and treatment modalities. Despite it being a major complaint, few studies have assessed pain as a primary or secondary outcome in these rare disorders, leading to inappropriate or ineffective pain management and reduced quality of life (QoL). In addition, studies into the pathomechanisms underlying chronic pain in these conditions, are very scarce. Intriguingly several studies have recently unveiled a role for the ECM in the maintenance and the chronification of pain.

Among the HCTD, chronic pain is strikingly present in individuals suffering from Ehlers-Danlos Syndrome (EDS).
EDS is characterized by hyperextensible skin, hypermobile joints and generalized soft connective tissue fragility and, like osteogenesis imperfecta, caused by genetic defects in fibrillar collagen biosynthesis and/or organization.

We systematically investigated pain and somatosensory characteristics in a cohort of individuals with molecularly confirmed classical EDS (cEDS) (mutations in either COL5A1, COL5A2 or COL1A1) and in age- and sex matched controls, and observed chronic pain associated with worse health-related QoL and altered somatosensory perception in the cEDS cohort compared to controls.

In a col5a1+/− mouse model for cEDS, we also observed mechanical, but not thermal, allodynia and abnormal cutaneous innervation in the glabrous skin of the hindpaw. We performed bulk RNA sequencing on the dorsal root ganglia from wildtype and col5a1+/− mice, and detected sex-dependent gene expression differences, with inflammatory pathways and cell adhesion pathways being disturbed in male mice while neuronal alterations were observed in female mice.

Together, our studies provide interesting insights on the possible role of the ECM in the development and persistence of pain.

**Disclosure**
None declared

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**IS21**

**How we work in the MDT pain clinic: Bristol and Oslo**

**Jacqui Clinch**
Bristol Royal Hospital for Children, Bristol, United Kingdom

An overview of the interdisciplinary pain team approach, both in the dedicated OI setting and also in our national (UK) residential pain service.

**Disclosure**
None declared
IS22

Modalities to deal with pain caused by fractures and surgeries

Cathy Raggio

Planning for surgical procedures involves an accurate assessment of the expected pain-intensity and duration. In general, discussions should involve the surgeon and anesthesiologist with the patient to plan and have correct expectations. Multimodal approaches to pain are generally used. The importance of pre-op planning both from a maintenance and surgery specific pre-Hab prospective will be key. Acute fracture pain treatment will be reviewed for pediatric and adult populations. Recommendations from NCBI will be discussed.

Disclosure
None declared

IS23

Pharmacological pain management

Richard Keen
Royal National Orthopaedic Hospital, Stanmore, United Kingdom

Pain is a common symptom in adults with osteogenesis imperfecta (OI) and can have a significant impact on the daily life, mobility, participation, work and social relationships of patients.

Chronic musculoskeletal pain associated with OI can be secondary to a number of factors including deformities after fractures, impaired muscle function and fatigue, premature degenerative joint disease and pain related to joint hypermobility. Some patients may also have features consistent with primary chronic pain which can be exacerbated by fatigue and poor sleep pattern.

The World Health Organization (WHO) created a practical pain ladder diagram in 1986 to help guide clinicians treating cancer pain throughout the world. This has since been adopted outside of oncology as a simple tool to manage pain. The WHO Pain Ladder utilises progressive steps of 1) anti-inflammatory agents, 2) weak opioids, and 3) strong opioids.
Some patients with OI may have chronic, primary pain and for these patients use of antidepressants should be considered. In addition, some patients report reduction in pain symptoms associated with intravenous bisphosphonate treatment.

This presentation will review the classes of drugs that are commonly used for pain management. The potential impact of these drugs on adult bone health will be explored. This will aid prescribers as they are informed about the benefits and risks of these treatments in adults with OI.

Disclosure
None declared

IS24

Therapeutic pain management principles in OI

Sophie Barlow
Royal National Orthopaedic Hospital, Stanmore, United Kingdom

Osteogenesis Imperfecta (OI) affects approximately 1 in every 10,000 people. This patient cohort has a wide range of healthcare needs, including musculoskeletal disorders, chronic pain and mental health issues which impact on quality of life. The NICE guidelines (UK) highlight the importance of healthcare professionals gaining an understanding of how a person’s pain affects their life, including their work and leisure time, relationships with family and friends, and sleep.

A tertiary service offers an individualised multidisciplinary approach to managing pain. Patients are assessed and treated by an occupational therapist, a physiotherapist and a psychologist. The programme focuses on how to gain occupational balance, explore physical activity as well as learning coping strategies to support with living well alongside pain. The course includes educational and practical sessions during a three week admission and are delivered both in groups as well as individual sessions.

Patients set goals at the end of the three weeks to enable them to continue with behavioural change and are followed up at three months and one year. Due to the nature of OI, a more bespoke programme is sometimes indicated and this can be facilitated with the support of the MDT.

Disclosure
None declared
IS25

Physical activity in OI: A good natural medicine for pain and health in OI

Miguel Rodríguez Molina
AHUCE, Madrid, Spain

Background
Nociception in OI comes from a mixture of low bone mineral density, lack of mobility, lack of strength, joint hyperlaxity, fatigue, bad alignment of the load axis of bones, early degeneration, body deformities and probably some unlucky consequences of fractures and surgery. Pain, as a subjective, multifactorial and personal sensation, requires a personal and multidisciplinary approach. Not all patients with OI are affected by those clinical problems in the same way, intensity, duration, time of their lives, etc. Improving those clinical features means decreasing painful sensations.

Objectives
Explain how and why physical exercise can improve these conditions and modulate pain.

Methods
State of the art and explanation of pain in OI and its possibility of treatment through some particular clinical cases where pain is caused by: low BMD, lack of mobility, lack of strength, joint hyperlaxity, fatigue, bad alignment of the load axis of bones, early degeneration, body deformities and some unlucky consequences of fractures or surgery. Physiotherapy and adapted physical activity is scheduled for each case and evaluated.

Results
Different investigations point favourably to the benefits of physical exercise in these different clinical situations, which will result in a long-term benefit of pain. We put it in contrast with the different clinical cases and evaluate each one. Conclusions: Regular exercise scheduled and adapted to each person with OI should be one of the main tools to improve pain and quality of life.

Disclosure
The author works as physiotherapist for AHUCE (Spanish national OI association).
**IS26**

*Creation of educational resources for children with osteogenesis imperfecta experiencing acute and chronic pain*

**Kelly Thorstad**

In an effort to address the paucity of pain educational resources available, our team at Shriners Hospitals for Children®-Canada has developed several tools and platforms to assess, educate, evaluate and share about pain in OI. These resources strive to target the multidimensional components of pain, and tackle the different pain experiences encountered by children in the hospital, home and community settings.

These resources include: OI Transfer Summary Tool (Carrier et al., 2018), Sisom OI (Siedlikowski et al., 2020), the Good2Go Passport (Jeong et al., 2019), Teens OI (Tsimicalis et al., 2023), OI Colour, OI Learn, The Dream Machine, and the PICH2GO Coping Kit, and others.

These resources are in varying stages of production and implementation and are available to the OI community for use and sharing.

**Disclosure**

None declared

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**IS27**

*A toolbox for pain in OI: empowering patients to take charge of their health*

**Ariane Kwiet**

*TRS National Resource Center for Rare Disorders, Norway*

People with Osteogenesis Imperfecta can have different types of pain during their life. They can experience both acute pain in relation to surgery or fracture and chronic pain of different categories.

Pain management can be difficult for the patient. The patient can feel alone with the pain and feel the need for more information about pain itself, pain treatment and pain coping.
There are many different toolboxes published for this purpose but none is designed explicitly for patients with Osteogenesis Imperfecta. Many of these toolboxes are relevant regardless the cause of pain, but there may be some features that these toolboxes do not cover that are important for patients with Osteogenesis Imperfecta.

That’s why the Osteogenesis Imperfecta Federation Europe (OIFE) and the OI Foundation (OIF) have come up with the idea to develop a toolbox designed for OI patients with pain.

In order to get this process started, there is a need to answer several questions:

1. What are the differences between patients with pain and Osteogenesis Imperfecta and patients with pain without Osteogenesis Imperfecta?
2. Is it possible to use an existing toolbox and add specific information for Osteogenesis Imperfecta or does it need a new toolbox? Which toolbox would be useful?
3. What is the target group for the toolbox? Children? Adults? Acute pain? Chronic pain? Where is the most urgent need?
4. What should be addressed in the toolbox? Understand pain? Communicate pain? Treat pain?
5. How should the toolbox be designed?
6. We want to discuss the needs, the design and the contents of such a toolbox and explore how we can get started with this process.

**Disclosure**

None declared
Sleep quality, excessive daytime sleepiness and increased risk for OSA in patients with osteogenesis imperfecta

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Objectives
In osteogenesis imperfecta (OI), beside the robust symptoms associated with locomotor system, symptoms may have detrimental effects on quality of life and sleep quality and promote development of various sleep disorders. One of the most common sleep disorders is obstructive sleep apnea (OSA), characterized by repetitive cessations of breathing during sleep and is associated with daytime sleepiness and an increased risk for cardiovascular and metabolic disorders. Recent research and data suggest a bidirectional causality between sleep apnea and disorders of bone metabolism. Such comorbidities, along with pain and fatigue, might contribute to the low sleep quality of OI patients. Thus, our aim was to assess the sleep quality, daytime sleepiness, and risk for OSA among patients with OI. We hypothesized that patients with OI have poor sleep quality, excessive daytime sleepiness and increased risk for OSA.

Methods
A total of 20 patients (11 males) with OI participated in this research, diagnosed with type 1 (25%), type 3 (40%), and type 4 (30%) OI, and a missing data regarding the type for 1 respondent. The average age was 26.4±19.4 years. We used Pittsburgh Sleep Quality Index (PSQI), Epworth Sleepiness Scale (ESS), STOP and STOP-Bang questionnaires, in an online format.

Results
The risk for OSA, according to STOP, was reported in 5 (25%) patients, whereas STOP-Bang revealed a risk for OSA in 8 (40%) patients. Patients reported an average PSQI value of 6.1±2.8, with 55% reporting values of PSQI>5, indicating low sleep quality. Average daytime sleepiness was 3.3±2.8, with just one patient reporting excessive daytime sleepiness.

Conclusion
In this group of OI patients, STOP-Bang more frequently recognized the risk for OSA than the STOP questionnaire. The frequency of risk was slightly higher when compared to our previously reported values in the general population of similar age.
Furthermore, low sleep quality was reported by the majority of respondents, which was not the case with excessive daytime sleepiness. Future research should aim for a similar study on a larger group preferably as case control comparisons addressing the postulated reduced sleep quality and elevated risk for OSA in the patients with OI.

Disclosure
None declared

S02

Evaluation of mobility associated symptoms in Argentinian patients with Osteogenesis Imperfecta. Application of the STEMS tool (Screening Tool for Everyday Mobility and Symptoms for Skeletal Dysplasias)

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Biomechanical alterations, such as short stature, limb incurvation and scoliosis affect mobility and may predispose to the presence of related symptoms such as pain and fatigue in Osteogenesis Imperfecta (OI). The Screening Tool for Everyday Mobility and Symptoms (STEMS) assesses mobility and associated symptomatology in individuals with skeletal dysplasias (SD) older than five years.

Objectives
To assess the presence of symptoms associated with mobility in everyday settings in people with OI using a Spanish version of the STEMS.

Methods
We performed a descriptive study in individuals with OI older than 5yo at the SD clinic at Garrahan Hospital, Argentina. Data collected included the severity of the condition, comorbidities, auxological, socio-environmental and therapeutic variables.

Using the STEMS the presence of pain and fatigue associated with mobility was evaluated. Additionally, pain and fatigue intensity were assessed using Spanish versions of validated scales. A pilot study evaluated STEMS comprehension and time required for completion.
Results
55 individuals with OI participated, of whom 30 had mild forms and 25 moderate and severe forms. Pain was present in 61.8%, fatigue in 85.5% and both symptoms in 52.7% of them. The presence of symptoms was more frequently associated with outdoor mobility. We found some associations, although not reaching statistical significance. The frequency of symptoms was associated with age and severity of the condition. The presence and intensity of symptoms were lower among those receiving bisphosphonates, doing physical therapy, and using mobility assistive devices. Having not appropriate schooling was more frequent among those who had symptoms. There was a positive correlation between the intensity of pain and fatigue, and between the intensity of symptoms with age, weight and body mass index. We have found some inconsistencies between the results of STEMS and the pain and fatigue intensity scales. STEMS was easy and quick to answer, the information collected was considered by the participants to be useful.

Conclusion
Using STEMS we found the presence of pain and fatigue associated with mobility in a high percentage of individuals with OI. These symptoms, usually naturalized, interfered with functioning, and were related to auxological and therapeutic variables.

Disclosure
None declared

S03

First results of using the Key4OI PROMIS questionnaires for measuring quality of life in children with OI, compared to children with other types of skeletal dysplasia

Marjolein Verhoef, Laura van der Horst, Wouter Nijhuis, Ralph Sakkers
UMCU, Utrecht, Netherlands

Objectives
The clinical manifestation of osteogenesis Imperfecta (OI) shows a wide variation. Therefore, care for patients with OI requires an interdisciplinary approach. The Key4OI international interdisciplinary working group developed a standard set of outcome measures focused on the needs and wishes of individuals with OI and their families, according to the ICHOM methodology. The aim is to gain a better understanding of the different aspects of quality of life of children with OI.
Method
In 2020 the national center of expertise for skeletal malformations of the University Medical Center in Utrecht (UMCU) started implementing the Key4OI outcome measures for children attending the clinic. In preparation of their visit to the expertise team, parents as well as children completed the questionnaires. In this study we will share the preliminary results of the PROMIS questionnaires for children aged 8 to 18 years on the domains Emotional distress-Depressive Symptoms, Emotional distress-Anxiety, Physical Function-Mobility, Physical Function–Upper Extremity, Pain Interference, Fatigue, and Peer Relationships. We applied the Computer Adaptive Test (CAT) version to limit the number of questions of the questionnaires. All participants in the study gave permission to use the data.

Results
The data is currently being processed and will be presented at the conference in June. A first comparison will be made of children with OI to children with other forms of skeletal dysplasia (e.g. achondroplasia, multiple epiphyseal dysplasia, spondyloepiphyseal dysplasia). Finally, challenges of implementing an (international) data set will be discussed.

Conclusion
Join our session at the conference to find out more.

Disclosure
None declared

S04

The European Registry for Rare Bone and Mineral Conditions (EuRR-Bone): collecting patient reported outcomes on osteogenesis imperfecta

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Introduction

The European Registries for Rare Bone and Mineral Conditions collect generic and condition-specific data elements using a Core Registry, a platform that is accessible to healthcare professionals and patients for data entry.

Methods

The EuRR-Bone Working Group on Osteogenesis Imperfecta (OI), a multidisciplinary team composed of healthcare professionals and patient representatives, worked together to develop a condition-specific dataset and issued recommendations on the use of Patient-Reported Outcome Measures (PROMs) in patients with OI.

Results

The final dataset included 34 Clinician-Reported Outcomes (CROs) covering domains such as anthropometry, genetic diagnosis, fractures, spine deformity, lung function, bone mineral density medical therapy and surgery. Twenty-two Patient-Reported Outcomes (PROs) were included in the final dataset. These outcomes include healthcare professionals involved regularly in the clinical care as well as those that are needed but unavailable. A section dedicated to medication consumption collects type, indication and frequency. Type of pain medication and frequency is also collected. Additionally, the Working Group recommended the use of EQ-5D for patients with OI which is a generic instrument used to measure health-related quality of life. It comprises 5 dimensions: pain or discomfort, mobility, self-care, usual activities and anxiety/depression. PROMIS health measures, specifically profiles, were also recommended by this and other Working Groups in EuRR-Bone.

However, its electronic use in addition to translations entail high costs. Since pain was a common interest of all the Working Groups we explored adding pain specific measures.
For this purpose, the Brief Pain Inventory - Short Form (BPI-SF) was added to the Core Registry. The BPI-SF is available and validated in multiple languages and provides 2 main scores, a pain severity score and a pain interference score. It additionally inquires about painful areas using a body map and the use of pain relief medication.

**Conclusion**

A better understanding of the impact of OI in health-related quality of life is needed. Longitudinal collection of CROs and PROs in the same platform will help fill in knowledge gaps in domains such as pain. Adding the standardised measures EQ-5D and BPI-SF can allow monitoring of changes in self-reported health status through time and in response to treatment.

**Disclosure**

Both project ‘777215 / EuRRECa’ (PI Faisal Ahmed) and project ‘946831 / EuRR-Bone’ (PI Natasha Appelman-Dijkstra) received funding from the European Union’s Health Programme.

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**S05**

*Pain and fractures in osteogenesis imperfecta: results from a survey of patients and caregivers*

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**Objective**

Osteogenesis Imperfecta (OI), also known as Brittle Bone Disease, is a rare genetic disorder commonly caused by variations in the formation of collagen, a major component of bone and other connective tissue. OI is associated with reduction in bone mass and increased bone fragility contributing to increased rates of fracture. Patients with OI can also have short stature, skeletal abnormalities, increased joint flexibility, muscle weakness, breathing problems, and hearing loss. The objective of this analysis was to examine patient- and caregiver-reported information to determine the impacts of OI on their lives, with a focus on fractures and pain.

**Methods**

Adults with OI and caregivers of children with OI were recruited through patient advocacy organizations. Participant eligibility was confirmed prior to them providing informed consent.
Participants completed an IRB-approved online survey regarding OI diagnosis, symptoms, fractures, and management. The survey is ongoing and results as of February 27, 2023 are presented herein.

Results
A total of 87 responses were received from 57 adult patients and 30 caregivers. Respondents were located in 16 countries, primarily in the United States (49/87, 56%), United Kingdom (14/87, 16%), and Spain (4/87, 5%). Participants reported diagnoses of OI Types I (40/87, 46%), III (13/87, 15%), IV (12/87, 14%), or Other/Unsure (22/87, 25%). Most (85/87, 98%) participants reported having had at least one fracture in their lifetime. When asked to rank the top 3 symptoms of OI that interfere most with their or their child’s life, both adults and caregivers frequently mentioned pain. Participants also frequently reported pain management as one of the top 3 most meaningful areas of improvement for an effective treatment for OI.

Conclusions
The survey results presented expand our understanding of the impacts of fractures and pain in adults and children living with OI. As additional survey responses are received, more areas of improvement in OI management may be identified.

Disclosure
All Authors: Employees, Stockholders: Ultragenyx Pharmaceutical Inc.

S06
Sleep parameters in adults with OI and their associations with pain and fatigue

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Objectives
To assess sleep parameters in adults with OI and identify their associations with pain and fatigue.
Methods
Thirty-five adults with OI (27 type I, 5 type III, 3 type IV; 54% females), mean age 44 years (±13.2), wore accelerometers (ActiGraph) for seven consecutive nights at the dominant wrist. Simultaneously, a diary was completed which assessed painful body area (Margolis pain diagram, higher scores indicate larger painful body area), pain severity (Multidimensional Pain Inventory, higher scores indicate higher pain severity), subjective sleep quality (self-designed questions, higher scores indicate lower sleep quality), sleepiness (Epworth Sleepiness Scale, higher scores indicate higher sleepiness levels) and fatigue severity (Checklist Individual Strength, higher scores indicate higher fatigue levels). Data are shown as median [interquartile range] or mean ± SD. Association (Spearman correlation) analyses and one-sample t-tests were performed.

Results
Median sleep efficiency was 90% [88;92], wake after sleep onsets (WASO) 45min [35;58], subjective sleep quality 5.6 [4.6;8.1] out of 18, and daytime sleepiness 3.9 [2.3;7.1] out of 24. Mean latency was 3 ± 2.6min and total sleep time 7h49 ± 46min. All sleep parameters indicated good sleep quality and no excessive sleepiness, except for WASO which was significantly higher (p<0.001) compared to norm values (<30min). Higher painful body area was associated with higher sleep efficiency (rs=0.432, p=0.015), lower WASO (rs=-0.408, p=0.023), lower subjective sleep quality (rs=0.359, p=0.043), and higher fatigue severity (rs=0.502, p=0.004). Higher pain severity was associated with lower subjective sleep quality (rs=0.432, p=0.014) and higher fatigue severity (rs=0.555, p=0.001). Finally, higher fatigue levels were associated with lower subjective sleep quality (rs=0.465, p=0.008) and higher daytime sleepiness (rs=0.415, p=0.020).

Conclusion
Adults with OI show higher amounts of time spent awake after initially falling asleep and before waking up, though without impact on their sleep efficiency or other objective sleep parameters. Higher extent and severity of their pain are both associated with higher fatigue levels and lower perceived sleep quality, though were not associated with impaired objective sleep parameters. Finally, higher sleepiness and lower perceived sleep quality were associated with higher fatigue levels.

Disclosure
None declared
Perceived dental care needs and concerns of individuals with osteogenesis imperfecta (OI)

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Objectives
Dental concerns are common in individuals living with OI, and up to 50%, depending on type, are thought to have Dentinogenesis Imperfecta (DI). There is limited evidence regarding the standard of care individuals in the UK receive. To investigate current issues and care experiences from the perspective of those living with OI, the Brittle Bone Society (BBS) distributed a survey in the UK and Ireland.

Methods
A survey was created in conjunction with the MAB, SAB and individuals with the condition, and distributed via the BBS social media platforms during Feb 2023. The survey explored 3 main themes looking at dental problems, dental concerns and accessing care both in the community and within dental hospitals.

Results
The survey had 110 respondents, 23 (21%) were 16 years and under, and 87 (79%) adults. The majority 77 (70%) reported having dental problems related to their OI, 48 (44%) stated they had DI and 32 (29%) were unsure if they had DI. Of 110 respondents 75 (68%) reported having concerns with appearance of their teeth, 49 (44%) had issues with their bite, 26 (23%) reported experiencing dental pain and 20 (18%) had pain in jaw.

The main dental issues in the community were: extractions 36 (33%), check ups 33 (30%), fillings 29 (26%). Reasons for not being able to access treatment included: 44 (40%) due to the lack of experience or dentist not feeling comfortable, 21 (19%) due to previously being on bisphosphonates and 11 individuals (10%) stated it was due to currently being on bisphosphonates. In comparison once referred to a Dental Hospital 59 (54%) stated they did not have issues with accessing treatment.

Conclusion
Dentists need to be more aware of the effects of both DI and effects of Bisphosphonates in relation to treatment to improve care to individuals with OI and more research is needed in this area.
Further research is also needed looking at the OI genotype / phenotype and a better understanding of pain / periodontal issues / and other dental anomalies.

Disclosure
None declared

S08

**Biological plates in the surgical treatment of imperfect osteogenesis**

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**Objectives**

It is known that screw plates are not a good option for the surgical treatment of long bones in patients with fragile bone, especially in osteogenesis imperfecta (OI). But sometimes solitary intramedullary nails do not get enough stability.

These are the cases, for example, of bone defects in the cortex or pseudoarthrosis of the diaphysis of long bones.

With this study we want to assess the effectiveness of these plates and disseminate whether it is the case, their use as a good therapeutic option in the fragile bone.

**Methods**

We reviewed total grafts as a biological plate that we have used in our hospital from the first in 2011 to December 2022, although the 74% of cases were used in last 5 years. We value base disease, surgical indication, its location, and evolution of the plate.

**Results**

We have used biological plate in 16 patients (23 cases). The base disease was OI in 17 cases, 4 fibrous dysplasia and 2 cases of congenital tibia pseudoarthrosis. 17 were femurs, (7 hips), 5 tibias and 1 humerus.

Regarding OI, 17 cases, 12 patients, in 9 the surgery was performed for nonunion of long bones, 2 for coxa vara, 5 fracture and 1 bone defect. 16 femurs (6 hips) and 1 humerus. The evolution of plate was in 6 cases osseointegration, in 2 it was reabsorbed and in 5 cases remained inert. 4 cases are pending evolution.
Conclusion
Biological plates are heterologous cortical sheets of bone. They act like metal plates but are fixed with wire and over time they can be integrated into the bone, eliminating the risk of fracture, and improving its mechanical situation.

Most of the surgical indications were non-union (they stabilize the focus and help consolidation), although sometimes the failure attributed to the biological plate may be due to the non-union and not the plate.

Disclosure
None declared

S09
Intramedullary canal sclerosis - a possible complication of prolonged bisphosphonate therapy in children with osteogenesis imperfecta

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Objectives
The pain is one of the biggest complaints of patients with osteogenesis imperfecta (OI). Successful treatment of the disease leads to satisfactory pain decreasing. Bisphosphonate (BP) intravenous infusions are a cornerstone of the treatment of OI. Prolonged BP therapy in case of significant deformity of the long bone will cause a problem during a surgery – sclerosis of intramedullary (IM) canal. The main goal of our study was to show the changes in long bone morphology which were connected with prolonged BP administration before the surgery.

Methods
The inclusion criteria for this study were patients without previous surgery, severe deformation of long bone segment (more then 50 degree of varus or procurvatum) and prolonged treatment with BP before the surgery. All radiographs were reviewed by single reviewer. IM sclerosis was measured on lateral radiographs because severe deformed segments had a specific “rib shape”.
Both ends points of sclerotic area were determined, and a whole surface of sclerotic area as well as percentage of sclerotic part was calculated. An angles of varus and procurvatum were measured also

**Results**
We found 17 segments (14 femurs and 3 tibias) in 9 patients which had some amount of IM sclerosis. All of included children were treated with intravenous infusions of pamidronate. The percentage of sclerosing part is decreasing till the fifteenth pamidronate infusion, after that we found its increasing. Obstacles during surgery were splitting of bone during drilling; drill breakage; “false route”; prolonged surgical time and increased blood losses. Our suggestions on how to avoid surgical difficulties are: precise preoperative planning, precise operative technique, many x-ray controls, gradual and patient reaming, reaming with surgical drill (instead of canulated drill) and excision of the bone block (triangular or trapezoidal shape).

**Conclusion**
The correct way of treatment of OI is to start with BP first, to prevent long bone IM sclerosis one should decide when to perform surgical long bone deformity correction. According to our results, the threshold for the operative treatment should be 15 cycles of BP infusions. Anticipating and avoiding difficulties and complications of surgical treatment leads to better results and consecutively adequately pain decreasing.

**Disclosure**
None declared.

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**S10**

*Pain and quality of life in patients with fibrous dysplasia/McCune Albright syndrome: a prospective follow up study*

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Objectives
This multicenter prospective observational study aimed to evaluate pain status in patients followed in the Fibrous dysplasia/McCune Albright (FD/MAS) care pathway in 2 tertiary clinics in the Netherlands.

Methods
243 patients (15-78 years) with FD/MAS (Leiden=217, Radboud=26) completed a validated questionnaire (Brief Pain Inventory) assessing pain severity score and Quality of Life (QoL) quantified by pain effect on functioning at baseline and after 1, 2 and 3 years of follow-up between 31 July 2018-1 March 2023 within the prospective FD/MAS study. Differences in pain scores were assessed between patients grouped by year of follow-up.

Results
193 patients were new referrals (79%), 162 females (66%). 110 patients (45.3%) had monostotic disease, of which 49 patients (45%) had isolated Craniofacial Fibrous Dysplasia (CFD), 95 patients (39.1%) had polyostotic disease (PFD), while 38 patients (15.6%) had McCune Albright syndrome. Patients reported improved scores in questionnaire’s function ratings from baseline until the 3rd year of follow-up (p<0.01). Females reported higher scores of maximal pain (4.9±2.8, p=0.019) and average pain (3.5±2.3, p=0.029). In women pain had a higher influence on conducting normal work (including householding) (2.9±2.9, p=0.041).

Patients with PFD had locomotion difficulties compared to those with isolated CFD (p=0.037), while there were no differences in pain scores between different subtypes of FD/MAS, meaning patients with monostotic and polyostotic disease had similar pain scores. 22 patients (11.3%) reported no pain at baseline and this remained throughout the study.

Pain treatment with NSAIDs, morphine, tramadol improved pain scores (p<0.01) and positively improved scores among all questioned areas (p≤0.001). FD-related treatment (bisphosphonates and Denosumab) improved scores of maximal pain (5.7±2.5 vs 4.4±2.8, p=0.03), minimal pain (7.5±0.7 vs 1.8±1.9, p=0.01), average pain (4.7±2.3 vs 3.3±2.4, p=0.016) and improved quality of life regarding general activity (4.8±2.7 vs 2.6±2.9, p=0.04), mood (3.6±2.8 vs 2±2.6, p=0.01), walking ability (4.4±3 vs 2.7±3.2, p=0.02), ability to conduct normal work (4.1±3.1 vs 2.4±2.9, p=0.008) and also improved social relationships (3±2.9 vs 1.6±2.5, p=0.03) and enjoyment of life (3.7±3.3 vs 1.7±2.4, p=0.01).

Conclusion
Pain is an important part of life of patients with FD/MAS independently of FDMAS subtype. Treatment, either with bone modifying agents or with analgesics improved pain scores and QoL significantly.
S11

**Working life with rare diagnosis**

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**Objectives**

To find out how people with rare skeletal diseases handled working life and their use of measures to enter and remain in paid work.

**Methods**

Semi structured focus group interviews of 14 adults employed in paid work with rare physical diseases registered at TRS competence centre.

Results as reported by the participants:

- Working life is of great importance. Participation in working life is more important than the percentage of full-time or size of salary. To achieve a life-work balance, many chose not to apply for the most demanding career choices or positions, even if they qualified.

- Varying capacity proved an added challenge in working life. When it’s impossible to know one’s own capacity until you get up in the morning, it is hard to estimate your workday and plan your tasks. It is also difficult to find a balance between the wanted contributions and the constraints that the diagnosis leads to.

- Invisible conditions lead to specific challenges. There are well-known measures available to overcome many of the practical obstacles in the work environment. On the other hand, when the diagnosis is invisible, or the capacity changes from one day to the next, there are less tools to overcome the challenges.

- When a diagnosis is rare and not familiar among employers and colleagues, it can lead to uncertainty. Lack of experience with rare diagnoses often led to a passive approach when handling work related issues.
To be able to communicate the implications of the diagnosis is essential for a well-functioning working life. A prerequisite for optimal contribution at work seems to be that employers and colleagues acknowledge those who live with a rare diagnosis as the ones who know best what kind of measures are helpful.

Conclusion
Few studies have investigated coping strategies and challenges of people with rare skeletal diseases in working life. Our results indicate that being able to participate in paid work and recognized for the skills are of great importance. At the same time acceptance and recognition of the limitations is necessary to be able to stay in the workforce over time.

Disclosure
None declared

S12
Pain rehabilitation of patients with a rare skeletal dysplasia – adapting to their special needs within existing rehabilitation programs
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Objectives
Patients with osteogenesis imperfecta and other skeletal dysplasias can have the need for multidisciplinary rehabilitation programs after surgery, because of chronic pain or other impairments. In Norway, they are referred to such programs like all other patients. Given the nature of a rare disease, they will rarely meet other patients with the same diagnosis. Additionally, the knowledge about skeletal dysplasias is low for most health care workers in the rehabilitation centers. Based on experiences from TRS, National Resource Center for Rare Disorders (TRS) and user organizations existing programs struggle to attend to the special needs of this group of patients.

The aim of this project is to evaluate the feasibility of a cooperation between an existing rehabilitation program and TRS to improve the quality of care for patients with rare skeletal dysplasias.
Methods
This is a feasibility study and a cooperation project between the rehabilitation center Unicare Jeløy and TRS. The patients with skeletal dysplasias will be organized as a subgroup within a larger group of patients with chronic pain conditions. They will generally receive the same individually adapted rehabilitation program. Experts from TRS will add educational interventions with disease specific context and give guidance about treatment decisions to the healthcare workers delivering the program. Feasibility will be evaluated with a mixed method approach.

Patients and health care workers will be invited to share their experiences through focus group interviews. Patient-related outcomes will be measured using PROMIS 57 for mapping pain, physical function, depression, fear, fatigue, sleep disturbance and social participation. Health related quality of life will be surveyed with EQ5D.

Results
Data will be analyzed and presented in a descriptive manner.

Conclusion
Results from this project will show whether a cooperation between existing rehabilitation programs and a National Resource center is feasible and improves the quality of care for patients with rare skeletal dysplasia.

Disclosure
None declared

S13
Use of medical care by individuals with osteogenesis imperfecta in the Netherlands for neurological and pain-related issues

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Objective
The aim of this project is to describe the use of medical care for neurological and pain-related problems in people with osteogenesis imperfecta (OI) in the Netherlands, and to compare this to the total population.
Methods
The data of 675 OI patients from the Amsterdam UMC Genome Database cohort was matched with records of the Statistics Netherlands (CBS) cohort. Patients were categorized based on their harbored pathogenic variant. For this project, we examined hospital admission data (total number of admissions and admissions in the neurological department) and medication data. We compared the results, when possible, between pathogenic variant type subgroups and to the total population.

Results
Between 2013 and 2019, there were 2133 admissions (both clinical and day-care), with 57 (2.7%) in the neurology department. A greater proportion of people with OI used medication compared to the total population. OI patients used more medication(s) from the ATC category ‘Nervous system medication’ which includes opioids, antiepileptic medication, antidepressants, and antimigraine medication compared to the general population.

Conclusion
Only 2.7% of hospital admissions were registered in the neurology department. Our data did not allow the identification of the diagnosis/problem for which the patients were admitted. Nervous system medication usage was higher for all age groups in the OI group. More studies are needed to clarify whether these were used for pain-related problems and psychiatric or neurological disorders.


Disclosure
None declared

S14
Transition and follow-up in adult OI
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Objectives
A lot of work has been done to gain knowledge about OI in children and meet obvious challenges in the young population. What happens as children get older?
Transition from childhood into adulthood is a critical time for the young and their families. Key information concerning health-related issues may get lost and follow-up may change significantly. A lot of questions are yet to be answered to offer a solid transition and well-founded clinical care for the adult OI population.

**Approach**

To improve transition, we have established a close cooperation between the pediatric center and the adult endocrine department who takes care of adults with OI.

An adult physician and an adult nurse from the endocrine department are invited to join the final visit for each young individual with OI approaching 18 years of age at the paediatric department. Special needs, recent fractures, pain, and other current health issues are discussed. At this consultation, the young patient and their relatives are introduced to the adult team, important health information is passed on, and the next appointment at the adult department is planned.

**Outcomes**

Paediatric clinical care at the hospital follows international guidelines. A decade ago, a large cross-sectional study at the same hospital, has investigated the adult OI population thoroughly. This knowledge has been used to prepare an OI-clinic one day per month, where adults with OI are seen by a team of doctors and nurses familiar with OI and expected challenges.

Before the appointment at the OI-clinic patients are suggested a number of topics they may like to discuss at the appointment. A semi-structured consultation based on this information is planned. There is a close cooperation with orthopedics, odontologists, radiologists, and other interns who are involved when necessary.

A RedCap database has been prepared for quality purposes to note all booked investigations, frequency of biochemistry, imaging controls, and use of other specialties.

**Implications / significance**

We hope that this structured approach towards young and adults with OI will help to create a safe clinical follow-up and at the same time lead to important knowledge concerning the adult OI population.

**Disclosure**

None declared
Pain and its biopsychosocial impact in children with skeletal dysplasias

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Objectives
To describe the pain experienced by children with skeletal dysplasias and to determine which biopsychosocial outcomes are associated with pain.

Methods
Participants aged 0-18 years with a confirmed or suspected skeletal dysplasia were recruited from the multidisciplinary clinic for children with skeletal dysplasias at the Children’s Hospital of Eastern Ontario in Ottawa, Canada. Parents/caregivers completed a series of validated questionnaires during a routine clinic visit to evaluate their child’s pain (current pain and worst pain in the previous week as assessed by a 10 cm visual analog scale on the Pediatric Pain Questionnaire), health-related quality of life (HRQoL), fatigue, pain interference, symptoms of depression and anxiety, sleep, peer relationships, and physical function.

As part of the physical function assessment, participants aged ≥6 years also completed single a two-legged jump (to assess lower body muscle power). We summarized our cohort and outcomes using descriptive statistics (mean/standard deviation [SD] or median/rage as appropriate) and tested for associations using Pearson’s correlations.

Results
Thirty-two participants (41% female) with a mean age of 11.8 (SD 4.1) years were included. Diagnoses included achondroplasia (9/32; 28%), osteogenesis imperfecta (type I, III or IV, 5/32; 16%), juvenile osteoporosis (3/32; 9%), and other skeletal dysplasias (15/32, 47%). Twenty (63%) parents/caregivers reported that their child experiences pain. Of those, median current pain and worst pain in the previous week levels were 2.1 (range 0.2-9.8) and 5.0 (range 0.2-100), respectively.
The top 5 biopsychosocial outcomes associated with current pain were pain interference ($r=0.73$, $p<0.0001$), HRQoL ($r=-0.56$, $p<0.0001$), peer relationships ($r=-0.51$, $p=0.004$), fatigue ($r=0.45$, $p=0.01$), and depressive symptoms ($r=0.38$, $p=0.03$). The top 5 biopsychosocial outcomes associated with worst pain the previous week were pain interference ($r=0.72$, $p<0.0001$), HRQoL ($r=-0.65$, $p<0.0001$), fatigue ($r=0.53$, $p=0.002$), peer relationships ($r=-0.52$, $p=0.002$), and weight-adjusted single 2-legged jump score ($r=-0.49$, $p=0.05$).

Conclusion
Pain is prevalent in children with skeletal dysplasias and is associated with increased pain interference, fatigue and depressive symptoms, as well as decreased HRQoL, peer relationships and muscle power. The directionality of these relationships should be explored to determine appropriate targets for future interventions to help prevent and treat pain in children with skeletal dysplasias.

Disclosure
Leanne Ward reports study grants to institution from Ascendis, Catabasis, PTC Therapeutics, ReveraGen, Ultragenyx and Amgen, and consultancy fees to institution from Ipsen, Santhera, Amgen, Alexion and Ultragenyx. Frank Rauch reports study grants to institution from PreciThera Inc, Ultragenyx Inc and Catabasis. Marie-Eve Robinson declares study grants to institution from Ascendis Biopharma and Ipsen Biopharmaceuticals and consultancy fees from Ipsen Biopharmaceuticals and Ultragenyx.

S16

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

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Background
Disturbed muscle (strength, mass, and metabolism) and bone (mass, geometry, strength, microarchitecture, and metabolism) parameters have been shown in osteogenesis imperfecta (OI). However, physiotherapy targeting these parameters, including high-impact or high-intensity exercises, entails high fracture risk. We hypothesize that low-intensity training with blood floMarw restriction (BFR), which has proven to safely benefit bone health and muscle growth in healthy and clinical adult populations, could safely target muscle and bone parameters in OI.

Objectives
To evaluate the effect of low-intensity muscle strength training (LI-MST) with BFR on bone and muscle parameters, pain, and fatigue in adults with OI type I.

Methods
First, questionnaires and interviews will evaluate feasibility, acceptability, and safety (symptoms and adverse events) of LI-MST with BFR in 10 adults (18-40 years old, 5 women, 5 men) with OI type I, in a pilot study at Ghent University Hospital (Belgium) in 2022-2023.

In the subsequent randomized controlled clinical trial (RCT), based on power calculations (effect size estimate f=0.4, with 95% power, α=0.05 and 20% drop-out), we aim to recruit 75 adult men and women with OI type I, aged between 18 and 40 years old, in several countries. Exclusion criteria include pregnancy or <6 months postpartum, deep vein thrombosis, acute fractures, edema or recent surgery in arms or legs, and cardiovascular, respiratory or neuromuscular diseases or other contraindications to perform exercise. Participants will be randomly assigned (block randomisation) into LI-MST with BFR, LI-MST without BFR, or a control group (standard care).

The exercise group will perform home-based exercises 2-3 times a week for 12 weeks. Training intensity will be set at 20% of 1-repetition maximum (1RM) and reset every 4 weeks for increasing intensity. In the BFR group, pressure will be applied by an inflatable cuff at the most proximal site of the arm or leg, inducing venous occlusion and partial arterial occlusion. Muscle and bone parameters will be evaluated by peripheral quantitative tomography, dual-energy x-ray absorptiometry, 1RM measurements, and blood samples, while pain and fatigue will be measured by questionnaires, at baseline, after 12 weeks (intervention), 6 months (follow-up 1) and 12 months (follow-up 2).

Disclosure
None declared
Impact of advanced therapy on pain in OI

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Objectives
Mesenchymal stem cells (MSCs), as the progenitors of the osteoblasts, the main type I collagen secreting cell type in the bone, have been proposed and tested as an innovative therapy for OI with promising but transient outcomes. Several MSCs infusions could overcome the short-term beneficial effect of MSCs therapy several infusions.

Methods
We have performed a phase I cell therapy multicenter clinical trial in Spain (code: TERCELOI), to evaluate the safety and therapeutic potential of five reiterative histocompatible bone marrow mesenchymal stem cells infusions, along 2,5 years plus two follow-up years, in two non-immunosuppressed pediatric patients affected by severe and moderate OI. HLA-haploidentical MSCs were obtained from bone marrow aspirated donated by healthy siblings. In addition to the clinical parameters evaluated, the host response to MSCs was studied by analyzing the sera from TERCELOI OI patients, collected before, during and after the cell therapy. Even more, the quality of life was evaluated through the validated PedsQL questionnaire consisting of 23 items in four functioning areas; physical, emotional, social and scholastic. Pain reported by patients and their respective parents, was one of the items evaluated in the physical area.
Results
TERCELOI resulted in a decrease in the number of fractures, an improvement in bone parameters and increased mobility. The molecular studies indicated a pro-osteogenic paracrine response elicited by the cell treatment. Even more, from the beginning of cell therapy, both patients and their respective parents reported improvements in all the parameters interrogated in the quality of life questionnaire, including pain.

Conclusions
TERCELOI has demonstrated the safety and therapeutic potential of the reiterative stem cells infusions in pediatric OI patients. The effectiveness of reiterative infusions in OI patients, supported by the clinical and molecular findings, was reflected in improvement in the quality of life, being the pain relief a paramount parameter.

Funding information
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Disclosure
None declared

S18

Learning through film: contributing to a balanced life with OI

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Background
It is important for people living with osteogenesis imperfecta (OI), their relatives and the wider community, to have relevant knowledge about the diagnosis, and how the diagnosis should be handled. Over the last six years, TRS National Resource Centre for Rare Disorders in Norway has supplemented written information about OI with films.

Objective
To increase awareness, and spread knowledge about OI to health professionals, educators, caregivers and persons living with OI in a more accessible way.
Method
Using film to educate and inspire in order to more effectively convey important information, and thereby to contribute to increased quality of life for people living with OI. The films should also be subtitled in English to reach a larger international audience. When producing the films we always work together as a team with specialists on OI, communication and persons with the diagnosis and sometimes their families. In this way, we secure that information pertinent to the audience is included, and that the stories are told, in a way that captures the attention and educates.

Results, the films:
- Rehabilitation after surgery and fractures
- Being young and living with OI
- Handling infants with OI
- Choosing the right wheelchair
- What happens at the OI Clinic

Conclusion
The distribution of the films is important to make sure the right people know about the content and have access to it. We therefore cooperate with patient organizations in spreading the films, and in making sure to be available in a diverse array of channels. In addition, we promote our films in a context making it possible for the viewer to find more detailed information about OI on our website.

Disclosure
None declared

S19

Chronic pain and fatigue in patients with multiple osteochondromas and Ollier disease: A systematic review

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TRS National Resource Centre for Rare Disorders, Sunnaas Rehabilitation Hospital, Bjørnemyr, Norway

Objectives
Many patients with rare diseases suffer from chronic pain and fatigue, with consequences for quality of life.
In the last years, several review articles have been written on this topic e.g. in Marfan syndrome, in osteogenesis imperfecta, in arthrogryposis multiplex congenita and on pain phenotypes in several rare diseases. A current scoping review on fatigue in rare diseases underline the need for more research on fatigue in rare diseases.

Here we present an ongoing review on chronic pain and fatigue in patients with multiple osteochondromas (MO) and Ollier disease (OD), rare diseases that feature benign tumors in the skeletal bones. Common complications are bone deformity, restricted joint motion, interference with skeletal growth and risk for malign transformation. A large part of the literature focuses on genetics or surgical interventions. Recently, however, some studies suggested that chronic pain and fatigue is a problem in these diseases.

This systematic review has three main goals: a) present existing research on chronic pain and fatigue in MO and OD patients, b) make clinicians aware of current knowledge on fatigue and chronic pain c) identify gaps and topics for future research.

Methods
We conducted a systematic search in the following databases: MEDLINE (Ovid), Embase (Ovid), APA PsycInfo (Ovid), CINAHL (EBSCO), Science Citation Index Expanded (Web of Science). We will include studies examining children and/ or adults with either MO or OD and where chronic pain and/ or fatigue is part of the assessment.

Results
We found 676 studies. After screening abstract and title, we excluded 492 studies. We are in the process of full-text reading now. From the included studies, we will extract information on study background, patients, pain and fatigue (prevalence, type, intensity, duration, treatment and assessment tools). We will also assess study quality and focus on future research targets.

Conclusion
We use the method of systematic review to present the current knowledge base on chronic pain and fatigue in patients with MO and OD in a systematic way. Results of this review can be used in guidelines and as a basis for future clinical studies.

Disclosure
None declared
The importance of nail follow up: about a case

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Objectives
Intramedullary telescopic nails (ITN) are the best option for the treatment of fractures and deformities of the long bones in osteogenesis imperfecta (OI). The nail used is important, but more important is correct nail placement during surgery and frequent monitoring of progress after surgery.

The object of this work is to transmit the idea of the importance of monitoring patients with intramedullary nails to avoid complications that can occur.

Methods
We reviewed the clinical case of a patient, a 12-year-old woman. She came to our hospital in May 2022. We started drug treatment and after two cycles we performed the first surgery. She had been surgically treated in her country for both femurs and both tibias.

Results
In clinical examination it appears to be a type III. She has never walked. She presents a very accentuated antecurvatum deformity in the right tibia and antecurvatum and varus deformity of the left femur.

The radiographs confirm clinical appearance and also the presence of an intramedullary nail in the right femur and left tibia and a very high risk of fracture induced by the nails in both bones.

The surgery to try to put our patient on his feet was much more complex in the bones with nails. But it was necessary to change them. In the bipedal position, the bone would break because the point of the nail was driving into the cortex of the bone from the inside and at that point it was curved and very fine.

For the girl to gain the bipedal position, it was necessary to operate on her and remove the nails... even though it was very difficult and complicated.

Conclusions
These nails are made of steel. They must also telescope... But they are inside a very fragile bone: they disengage, they do not telescope and bone bends on the nail...
Frequent clinical and radiographic monitoring is very important. It allows timely detection of complications associated with ITN in OI and small surgical gestures carried out early can avoid extreme situations.

**Disclosure**
None declared

**S21**

*Balancing function with pain in a child with type V OI*

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*Hospital for Special Surgery, New York City, USA*

**Purpose**
Musculoskeletal pain is a frequent report by children with all types of Osteogenesis Imperfecta (OI). Assessment of pain-related disability and consideration of functional impairment are important factors that impact physical therapy evaluation, intervention, and progress toward child-centered goals.

This presentation aims to explore the objective and outcome measures used with children with OI to assess progress in an outpatient pediatric physical therapy setting. This presentation aims to present a case report of the physical therapy management of a child with OI type V and illustrate the use of these measures.

**Description**
Children with all types of OI may report pain related to an acute fracture and chronic non-fracture. Most children with OI report moderate to severe pain associated with acute fracture and less intense non-fracture chronic pain. Assessment of pain-related disability is important when evaluating and treating pediatric patients with acute and chronic pain. Children with OI report that their activities of daily living and participation are affected by fracture and non-fracture pain. This presentation uses objective pain measures specific to function at evaluation, treatment, and discharge. This presentation aims to review the pain-related objective measures utilized for a child with OI type V during acute fracture and non-fracture episodes of care to follow progress in PT and monitor any changes in functional status.
Summary of Use
Function-specific patient-reported outcome measures such as the Functional Disability Inventory, the Patient Specific Functional Scale, and Faces Pain Scale-Revised, are objective measures of pain-related disability, functional impairment, and pain intensity used at evaluation, during treatment, at discharge to monitor progress, to modify physical therapy, and to guide referrals to other health professionals as appropriate. These objective measures are patient-reported and help the therapists develop meaningful goals with the child and caregiver.

Importance to Members
Children with OI of all types report musculoskeletal pain, often in the absence of acute fracture or with analgesics. Pediatric physical and occupational therapists working with children with OI must understand the impact of pain-related disability and utilize appropriate assessment tools to identify pain and functional impairment throughout the therapy episode of care. A therapist’s understanding of the assessment tools will guide the development of therapy programs with a plan to get the children back to the activities that are important to them and ultimately want to participate.

Disclosure
None declared

S22
Abstract withdrawn

S23
Root resorption and dental maturity in children with osteogenesis imperfecta medicated with bisphosphonates
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Osteogenesis imperfecta (OI) is a tissue disorder characterised by repeated fractures and skeletal disorders. Bisphosphonate (BP) therapy is currently the gold standard in treating OI. Tooth development (TD) and root resorption (RR) take place at the expense of clastic cells. Administering BP during the growth and development period could alter these physiological processes.
Goals
To study the TD of permanent premolars and the RR of primary molars in children with osteogenesis imperfecta who are medicated with BP. To compare the results to a control group sample. Material and Methods Two groups were studied. A study group (n=26) of boys with OI who are medicated with BP and a control group (n=395), matched by age and gender. They were classified by age into two groups, one group aged 6-8.9 years and the other 9-11.9 years. Panoramic X-rays were used to measure root resorption of 7.4 and 7.5 using the Haavikko method, while the Demirjian method was used to establish tooth development of 3.4 and 3.5. The Mann-Whitney test was used, and statistical significance was considered at p<0.05.

Results
The mean age of the 421 participants was 9.21 years; according to gender, 52.5% were boys (221) and 47.5% were girls (200). RR of 7.4 and 7.5 and TD of 3.4 and 3.5 in children with OI is lower than the control group p<0.05.

Conclusions
Children with OI who are medicated with BP have less premolar tooth development and less root resorption of primary molars. However, the difference is not clear if studied by age subgroups

Disclosure
None declared
Eva Åström

Eva Åström is senior consultant at the Neuropediatrics at Karolinska University hospital and has a PhD at Karolinska Institutet in Stockholm, Sweden “Bisphosphonate treatment to children and adolescents with Osteogenesis imperfecta”. She has been assessing and treating children with Osteogenesis Imperfecta (OI) since 1991.

She started the first intravenous Pamidronate treatment in OI 1991 and reported the first case of intravenous Pamidronate treatment at the 5th International Conference on Osteogenesis Imperfecta in Oxford 1993. She is the leader of a Swedish pediatric multi-professional team for assessment and treatment of children and adolescents with OI (from 2023 licensed by the National Board of Health and Welfare to be Sweden’s only center for national specialized medical care for pediatric OI) and is main Principal Investigator for BOOSTB4, an international multicenter trial lead by Cecilia Götherström, to evaluate the clinical effect of early mesenchymal stem cell transplantation for the treatment of severe OI.

Sophie Barlow

Graduated with Bsc Hons degree from St George’s University of London, 2008. Having completed junior rotations at the Lister hospital in Stevenage she joined the Royal national Orthopaedic Hospital in 2011. She specialised in pain management and rehabilitation in 2013 and has been working alongside the multidisciplinary team working with adults with Osteogenesis Imperfecta for three years.
Michael B Bober

Dr Michael B Bober is a pediatrician and geneticist who directs the Skeletal Dysplasia Program at the Alfred I duPont Hospital for Children in Wilmington, DE. He is a Professor of Pediatrics at Thomas Jefferson University’s Stanley Kimmel Medical College. Dr Bober completed a combined MD/PhD program in Biomedical Engineering at Tulane University.

His dissertation research focused on the genetic response of bone to mechanical and hormonal stimulation. He then went on to complete a Pediatrics Residency at Tulane University and a Medical Genetics Residency and Fellowship at Johns Hopkins University. He is a board certified in Pediatrics, Clinical Genetics and Molecular Genetics. Clinically, his practice is exclusively focused on the diagnosis and management of children with skeletal dysplasia and osteogenesis imperfecta. Dr Bober has a special interest in intensive care during the neonatal period.

Alison Boyce

Alison Boyce, MD is a pediatric endocrinologist and Chief of the Metabolic Bone Disorders Unit in the National Institute of Dental and Craniofacial Research, National Institutes of Health. The goal of her work is to enhance health and well-being for children with skeletal disorders by developing novel tools and treatments informed by pathophysiologic studies. She leads investigations in fibrous dysplasia/McCune-Albright syndrome, a rare disease affecting the bone and endocrine systems, and has characterized many aspects of this disorder and its treatment.

Dr Boyce is a faculty member in the NIH Endocrinology fellowship training program. She is a member of the Fibrous Dysplasia/McCune-Albright Syndrome International Consortium and serves as Chair of the Medical Advisory Committee to the FD/MAS Alliance.
Karen Braitmayer

Karen L Braitmayer, FAIA is the founder and managing principal of Studio Pacifica, an accessibility consulting firm in Seattle. She and her team provide consulting services to local governments, school districts, architects, engineers, companies and individuals concerned with complying with Federal laws and State codes, as well as simply creating spaces that work for the unique needs of individual users.

Karen also leads presentations and workshops around the country to further educate professionals about codes, standards, and inclusion. Early in her career, it occurred to Karen that as an architect and a wheelchair user, it was possible for her to make a unique contribution to the field. Her professional focus on accessibility and her advocacy efforts for inclusion have certainly done that!

As a registered architect, Karen was admitted to the prestigious College of Fellows by the American Institute of Architects (AIA). In 2010 she was appointed by President Barack Obama to the United States Access Board, a policy position that she still holds today. Most recently she was awarded the 2019 Whitney M. Young Jr. award by the American Institute of Architects in recognition of her leadership in civil rights for people with disabilities, social sustainability, public policy and universal design. In addition, Karen was named 2019 Person of the Year by New Mobility Magazine. Karen is also an active volunteer and non-profit board member.

Meryl Chambers

Meryl is Chair and Coordinator of Soft Bones UK Hypophosphatasia Foundation. Meryl is married with two children, Maddox 9 and Ruby 7. Meryl studied Audiology (Hearing) at the University of Southampton and qualified in 2009. Since then, she has been working as an Audiologist across both the private and NHS sector. Her current role is in the NHS working as a paediatric audiologist and bone anchored hearing aid specialist.
Meryl became involved in the Hypophosphatasia community shortly after her son was diagnosed with the condition in 2014 when her son Maddox was just over a week old. Meryl sat on the panel as a patient representative for the NICE Highly Specialised Technologies Programme meeting for the drug Strensiq for use with Hypophosphatasia, which has now been recommended and approved by NHS England. It was shortly after this Meryl became involved with setting up Soft Bones UK, recognising the need for greater support for HPP families through sharing experiences, resources and contacts.

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**Jacqui Clinch**

Dr Jacqui Clinch is a consultant in child, adolescent and young adult rheumatology and rare bone disease at the Bristol Royal Hospital for Children, and Medical Lead for the National Adolescent and Young Adult Chronic Pain Service (Bath Centre for Pain Services).

Jacqui is on several national/international boards addressing aspects of inflammatory and non-inflammatory musculoskeletal disease, pain and osteoporosis in young people. She has written/co-written national/international reports and guidelines (for organisations including WHO, NHSE, NICE, CRG, RCPCH). Currently she is Vice President of the British Soc of Rheumatology, and Chair of NHSE MSK child and young person workstream.

Academically and educationally she is widely published with areas of interest including child and adolescent chronic pain conditions (disease and non-disease related), hypermobility, paediatric arthritis, parenting in chronic childhood disease and rare bone diseases. Specific recent interests include interventions in childhood MSK disease and pain, diagnostic uncertainty and impact of this in paediatricians, technology evaluating adolescent drug adherence and impact of MSK pain on young people and families. Politically she is committed to striving for equity of access to excellent care for all young people with MSK conditions across the UK.

Outside work she enjoys an active family life, running, squash, sloe gin and chickens.
Marie Coussens

Marie Coussens, PT, studied physiotherapy and specialized in internal diseases at Ghent University, Belgium. Currently, she is working as a PhD student at the department of Rehabilitation sciences and Physiotherapy of Ghent University, where her research focuses on individuals with hereditary connective tissue disorders. In collaboration with Macquarie University (Sydney, Australia), her work spans from the identification of the needs and determinants of reduced physical activity, muscle weakness and joint hypermobility, to altered bone parameters in individuals with Osteogenesis Imperfecta, hypermobile Ehlers-Danlos syndrome and Hypermobility Spectrum Disorder. Besides research, she teaches into the physiotherapy program at Ghent University and is involved in the Center for Medical Genetics of the University Hospital where she worked clinically with patients with heritable connective tissue disorders.

Marie Fahlberg

Marie H Fahlberg, Healthmotivator and 24 years of experience of fear and pain as a mother to a son living with the rare diagnosis Fibrodysplasia Ossificans Progressiva (FOP). She is an entrepreneur and founded the Swedish FOP association in 2004 and is now back as chairman after a few years’ break from board work, the association has members from the Nordic countries, her voluntary involvement for FOP is on a global level. Privately, she built up and runs the website fopsverige.se, a platform with information and continuing education about FOP. She is a licensed lifestyle and health coach, yoga instructor and body awareness guide with an extra interest in nutrition, breathing, movement, pain and how we ourselves can influence our well-being and simplify our lives while undergoing painful experiences in life. Website
**Kis Holm Laursen**

Kis Holm Laursen (57) Occupational Therapist since 1993. Started own company Det Unikke Barn (The unique child) 7 years ago. Helps parents prioritize daily activity for themselves and the children in the family, to gain confidence and strength. Gives presentation about cooperation with the parents for professionals. Gives lectures for parents and facilitate exchange of experience about family life with a child with special needs. Gives lectures for grandparents too. Guidance counselor in the Danish OI foundation since 2000, board member of DFOI 1999 - 2017. 2020, in cooperation with Charlotte Jensen, psychologist, created an online course “Chronic pain in School children” for teachers, pedagogue and nurses. Courses, education: Theraplay step 1, Sensory Defensiveness – a comprehensive treatment, Soma 1+2 - Trauma Treatment, Trauma Treatment in children, Children with pain (The autonome nervesystem, pain evaluation and relief, cooperation with the parents). Mom to Marie, 26, with severe OI, [TedX talk in 2023](#).

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**Alex Ireland**

Alex Ireland is a Reader in Musculoskeletal Physiology in the Musculoskeletal Science and Sports Medicine Research Centre at Manchester Metropolitan University. Working with human participants from clinical and non-clinical populations, his research examines the effects of mechanical loading on bones and joints across lifespan from prenatal movements to exercise in old age. In addition, the influence of skeletal conditions such as osteogenesis imperfecta and X-linked hypophosphatemia (XLH) on neuromuscular health and function.
Richard Keen

Professor Richard Keen is a Consultant in Metabolic Bone Disease at the Royal National orthopaedic Hospital, London, UK. After training in rheumatology he developed his specialist expertise and interest in osteoporosis and other rarer bone conditions. His team offer clinical care to patients with osteogenesis imperfecta (OI), and they run a dedicated multidisciplinary clinic for adult patients. Professor Keen has also developed formal transition clinics for young people with OI who are moving their care from Great Ormond Street into adult services. He has been Principal Investigator for a number of clinical trials looking at novel treatments for OI. Professor Keen is also the Chairperson of the Scientific Advisory Board for the Brittle Bone Society.

AnnBett Kirkebaek

AnnBett Kirkebaek is a fitness coach, master instructor and team manager at FitnessX, Denmark. She received a bachelor’s degree in organizational leadership with a concentration in project management, coaching and innovation. AnnBett has Osteogenesis Imperfecta. AnnBett has 12 years of experience as a fitness instructor. During these years, she has found firsthand how physical exercise can help, support, and relieve pain for the fragile bones.

AnnBett published an article on: Pain’s impact on relationships and sexuality when suffering from brittle bones: “I found that physical exercise can provide pain relief for those with OI. But I also found that the conversation about the pain is crucial and sometimes even a better pain relief overall”.

She is currently giving lectures in Denmark based on the article. AnnBett is married and the mother of two girls aged 21 & 18 which the youngest also has OI.
Ariane Kwiet

Ariane Kwiet is a specialist in physical therapy and rehabilitation and a pain specialist. She worked many years in a pain clinic and in a rehabilitation department. The last 4 years she worked at the TRS National Resource Centre for Rare Disorders in Norway with different skeletal dysplasia diagnoses. She is particularly interested in pain treatment and how rehabilitation possibilities can be improved for these patients.

Liana la Forgia

Liana la Forgia was born in Italy but moved to England in 1992 where she graduated in History and English. Subsequently she qualified as a teacher and has taught Italian language for over 20 years whilst raising a family. She is also the mother of a patient affected by MO.

Since 2017 Liana has been a volunteer for A.C.A.R. Aps, the Italian Patient Association on Multiple Osteochondromas, Ollier disease and Maffucci Syndrome, all part of the ERN BOND network. Her role within A.C.A.R. Aps is to communicate with other European Patient Associations for the above diseases.

In April 2022 Liana completed a Master’s Degree in Patient Advocacy Management at the Università Cattolica in Rome, Italy, and belongs to the ALUMNI of the university. In September Liana became an ePAG in ERN BOND and since then became further involved in advocacy and participated in new projects aimed at supporting the patients she represents.
Lena Lande Wekre

Lena Lande Wekre, senior consultant at TRS National Resource Center for Rare Disorders, part of the Norwegian National Unit on Rare Disorders (NKSD) and situated at Sunnaas Rehabilitation Hospital, Oslo. She is working in the skeletal dysplasia team, and did her PhD in adults with osteogenesis imperfecta. She works with patients of all ages, from newborn to death, and all persons with a rare skeletal bone-disorder are welcome to register at the center. Has been working with rare disorders since 1999. Over the last decades she has been working on several projects. In the Steering Committees of the European Reference Network for Rare Bone disorders, ERN Bond, and for the global IMPACT study of OI. Member of the Medical Advisory boards for OIFE and NFOI (Norwegian Osteogenesis Imperfecta Association). In collaboration with the Orthopaedic division, Children’s Orthopaedics and Reconstructive Surgery, at Oslo University Hospital she has developed a National Registry for rare congenital bone disorders.

Katarina Lindahl

Katarina Lindahl graduated from medical school at Uppsala University in 2003. After internship in Sundsvall she returned to Uppsala University Hospital for residency in internal medicine and thereafter subspeciality in endocrinology. From 2006 she attended a PhD-program and defended her thesis “Osteogenesis Imperfecta – Genetic and Therapeutic studies” at the Department of Medical Sciences of Uppsala University in 2013. The dissertation focused on a large cohort of Swedish patients with OI, a rare high bone density variant in this cohort, common collagen variants and siRNA studies in cell cultures of human primary bone cells aiming at silencing the pathological collagen variant.

In 2017 Katarina decided to leave clinical work for a position as a clinical assessor at the Clinical Trial unit at the Swedish Medical Products Agency, specializing in assessing clinical trials and providing scientific advice to academia and industry. Katarina lives in Uppsala with her five children and partner.
Fransiska Malfait

Fransiska Malfait is associate professor affiliated with the Center for Medical Genetics (CMGG) at Ghent University Hospital and the Department for Biomolecular Medicine at Ghent University, Belgium. She is Head of Clinic at the CMGG where she leads the outpatient clinic. She is specialized in clinical and molecular aspects of heritable connective tissue disorders (HCTD), with her research work focusing on three main topics: (I) Unraveling the molecular basis of HCTD, and studying their natural history and genotype-phenotype correlations (main focus on the Ehlers-Danlos Syndromes); (II) elucidating molecular and physiological mechanisms underlying HCTD pathogenesis, using an integrated approach of in vitro and in vivo techniques, on tissue samples of humans and animal models (zebrafish, mice); and (III) studying prevalence, nature and pathophysiologic mechanisms of pain in Ehlers-Danlos Syndrome patients and in relevant animal models. Visit her website if you want to know more: www.malfaitlab.org.

Rubén Muñoz Cortés

Rubén Muñoz Cortés lives in Valencia (Spain) and has been a clinical psychologist at the Spanish Osteogenesis Imperfecta Foundation (Fundación AHUCE) since 2015. There he works mainly in clinical care and psychosocial research. Since 2022 he is PhD in Clinical and Health Psychology with research on chronic pain in adult population with osteogenesis imperfecta.
Samantha Prince

Dr Samantha Prince is a Principal at Wickenstones Ltd., a global consultancy who work with pharmaceutical clients to help bring new medicines to patients. Samantha holds a PhD in medical and molecular genetics following doctoral research on rare syndromic diabetes and her most recent career experience is within market access consultancy, working with small-scale, national, and global clients on a variety of evidence generation and value communication projects.

Samantha’s project involvement spans a wide range of therapeutic areas but she has a particular interest in rare inherited conditions. Over the last 3 years, Samantha has managed the IMPACT Survey project, an international collaboration supported by Mereo BioPharma aimed at gathering insights about life with OI from people with OI and their families. She has had the great pleasure of working closely with a Steering Committee of OI clinical experts and community representatives from around the world.

Cathleen Raggio

Cathleen L Raggio, M.D. specializes in general pediatric orthopedics, scoliosis and in the treatment of osteogenesis imperfecta and skeletal dysplasia and is the Co-Director of the Kathryn O. and Alan C. Greenberg Center for Skeletal Dysplasias at Hospital for Special Surgery. Dr Raggio completed her residency in orthopedic surgery at Hospital for Special Surgery and fellowship in pediatric orthopedic surgery at Alfred I. duPont Hospital for Children. Known worldwide throughout the medical community, Dr Raggio performs basic science and clinical research on the etiology of scoliosis, osteogenesis imperfecta, osteoporosis, and skeletal dysplasias. Dr Raggio has helped define the different types of OI, specifically type IX and the different drug treatment options using the mice models of OI. She has served as Co-Chair of the International Research Conference in Annapolis and has served on the subsequent organizing committees. Dr Raggio is currently on the OI Foundation’s Medical Advisory Council and has spoken at the biennial National Conferences.
Mercedes Rodriguez Celin

Dr Rodriguez Celin is originally from Argentina, where she studied Medicine at the University of Buenos Aires and did a 4-year residency in pediatrics. After finishing her training, she practiced pediatrics with a specialty in Growth and Development. She served as an attending physician at the Skeletal Dysplasia Clinic in the Garrahan Pediatric Referral Hospital in Buenos Aires for six years. Her focus at Garrahan was on clinical assistance and clinical research in Osteogenesis Imperfecta (OI) and other Skeletal Dysplasias. From 2017-2022, Dr Rodriguez Celin worked as a clinician-researcher at Shriners Hospital for Children, Chicago. She completed an HHS/NIDILRR postdoctoral Fellowship in Advanced Rehabilitation Research Training (ARRT) through a multidisciplinary program offered through Marquette University, the Medical College of Wisconsin (MCW), and Shriners Hospitals for Children, Chicago. During her years at Shriners Hospital, Dr Rodriguez Celin was involved in several multicenter studies to improve the knowledge about the functional, surgical, and pain status of patients with OI. In 2022 Dr Rodriguez Celin started her internship training in pediatrics at the University of Illinois at Chicago, and she will start her training as Medical Geneticist at MCW in July 2023.

Miguel Rodríguez Molina

Miguel Rodríguez Molina is the physiotherapy of AHUCE and Fundación AHUCE in Spain. He is specialized in neuro motor control, neurological diseases and rare diseases. In the last 8 years he has been working with bone dysplasias, specially with Osteogenesis Imperfecta and he is member of the Medical Advisory Board of the OIFE. He is PhD Candidate form University of Alcala, researching about OI and soft tissues. He combines his scientific activity with clinical practice in Getafe, Madrid and Valencia.
Inger-Margrethe Stavdal Paulsen

Inger-Margrethe Stavdal Paulsen has a degree in Education from Østfold University College in Norway. She works part time as an elementary school teacher, currently teaching third grade. Inger-Margrethe is chair of the Norwegian Osteogenesis Imperfecta Association (NFOI), a volunteer position she has had since 2016. She is chair of the advisory board of TRS National Resource Centre for Rare Disorders, a graduate of Eurordis Summer School on Medicines Research and Development and part of the Eurordis HTA Task Force. Inger-Margrethe has a particular interest in patient involvement, especially on the regulatory side.

Michael Stewart

Michael Stewart (he/him) is the Director of Education at the Osteogenesis Imperfecta Foundation (OIF). In this role, he manages educational initiatives for medical professionals and OI patient communities, plans events, and helps various OI-experts more effectively share their knowledge. His major responsibilities include OIF Regional Conferences, the Rare Bone Disease and OI TeleECHO Clinic Series, Brittle Bone Disorders Consortium (BBDC) Research Updates, and other education related initiatives. Before coming to the OIF, Michael was a public high school social-studies teacher in New York City.
Audun Stubhaug

Audun Stubhaug is professor of Anesthesiology and Pain Medicine at University of Oslo and until recently was Head of Department of Pain Management and Research at Oslo University Hospital, Norway. He has been combining clinical work and pain research for several decades. He is currently involved in research within pain epidemiology, as well as clinical studies and human experimental studies. He is engaged in developing clinical registers and the documentation of societal impact of pain and developing cost-effective treatments. He has published 200 articles and book chapters in the pain field and is currently president of the Norwegian IASP-chapter.

Kelly Thorstad

Kelly Thorstad is the Director of Nursing and Patient Care Services / Nurse Executive at the Shriners Hospitals for Children®- Canada in Montreal. Ms Thorstad obtained her MSc(A) in Nursing from McGill University in 2001, and later continued her education at the University of Ottawa to obtain her Primary Health Care Nurse Practitioner license in 2008. She has been a part of the Shriners Hospital team since 2002 and has adopted many roles over the years, in managerial and clinical positions. Ms Thorstad has led nursing education and research initiatives, implemented clinical process changes, facilitated the transition to a new hospital facility, and has led with her colleagues to establish best practices in care delivery. Ms Thorstad played a key role in the implementation of the FOCUSED™ professional practice model across the 22 site Shriners Hospitals for Children system, which ensures that patients and families remain at the center of clinical care delivery. As a Nurse Practitioner, Ms Thorstad has been involved in the clinical care of children with orthopedic needs and has had a research focus on the transition of young adults, and optimization of pain, anxiety, and coping management. Under Ms Thorstad’s direction, the focus of the Canadian Shriners Hospital Nursing Research team has been on evidence based clinical care for patients and families living with Osteogenesis Imperfecta.
Tenna Toft Sylvest

Tenna Toft Sylvest was born in 1981 and lives in Copenhagen, Denmark with her husband and twin daughters. Tenna was diagnosed with X-linked Hypophosphatemia (XLH) at around 18 months as the first in her family. One of her twin daughters has inherited XLH. Tenna has a master's degree in political science and works as a Senior IT Consultant in the Ministry of Foods, Agriculture and Fisheries in Denmark. In 2018 Tenna co-founded the Danish patient organisation for XLH where she has served as president since 2018. The Danish XLH organisation also includes patients with HPP. In 2017 Tenna helped establish the International XLH Alliance together with representatives from 5 other countries and currently serves as Co-Chair. Tenna has been a member of the ERN BOND ePAG group since 2019.

Rebecca Tvedt Skarberg

Rebecca Tvedt Skarberg was born in the U.S. and lives today in Oslo, Norway. Her personal experience living with a rare bone condition comes from being born with osteogenesis imperfecta (OI). Rebecca is a trained social worker with additional degrees in psychology and counselling. She has previously worked for the Norwegian National Welfare office (NAV). From 2014 to present date Rebecca has been working for the Norwegian National Advisory Unit on Rare Disorders (NKSD). Her focus has been on strengthening patient involvement, strategy and planning of services for people with rare disease, as well as working with international issues.

Rebecca started volunteering from an early age through the Norwegian OI Organization (NFOI) and The Norwegian Federation of Organizations of Disabled People (FFO). Rebecca has been engaged in international volunteer work through OIFE for many years. She was a part of the establishment of BOND ERN and has served as an Epag since ERNs were launched in 2017. Rebecca is a member of the steering committee in ERN BOND and has also contributed in many working groups.
She was co-lead in the EuRR-Bone registry WP5 from 2020-23, especially working on patient involvement. In 2019-2021 she served on the Rare2030 panel of experts actively advocating for a renewed European Action Plan on Rare Disease. Rebecca Tvedt Skarberg was elected to the EURORDIS Board of Directors in 2022. She has recently been appointed to the EU Disability Plattform representing EURORDIS. Rebecca is a proud recipient of the Black Pearl Volunteer Award 2020.

Rebecca has been personally invested in disability rights, equity and patient advocacy from an early age. This has led her to be engaged in many projects and arenas for people with OI, with a rare condition and also people with disabilities on a whole. Her aim is to use her experience and drive to help empower people and engage professionals to spread awareness and knowledge.

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**Ingunn Westerheim**

Ingunn Westerheim (48) has a Master in law from the University of Oslo, Norway. In addition to formal education, Ingunn belongs to the alumni of EURORDIS Summer School and is also a certified scrum master (project management).

She has worked 15 years as a legal advisor in the Norwegian Labour- and Welfare Directorate and five years as a political advisor in The Norwegian Federation of Organisations of Disabled People (FFO). She served as Board leader for the Norwegian Osteogenesis Imperfecta Association (NFOI) from 2001 to 2016.

She has served as the President of Osteogenesis Imperfecta Federation Europe (OIFE) since September 2015. She represents OIFE at the Council of Federation of EURORDIS. Ingunn is also a member of the National coordinating group for the Norwegian network for precision medicine and The Norwegian coordinating group for Norway’s participation in ERNs.
Andrew Wiese

Andrew Wiese is a Licensed Clinical Psychologist and assistant professor in the Menninger Department of Psychiatry and Behavioral Sciences at Baylor College of Medicine. Andrew completed his PhD in clinical psychology from the University of Missouri – Kansas City, and his undergraduate education from Michigan State University. His research and clinical interests include obsessive compulsive and related disorders, and anxiety disorders. More recently, he has begun examining the intersection between these groups of psychopathology and health conditions, including osteogenesis imperfecta.

Jacob Østergaard-Christensen Wittorff

Jacob Østergaard-Christensen Wittorff (41) is a Danish tech journalist with a bachelor’s degree in social science and a master’s degree in journalism and communication from Roskilde University. Since 2008, Jacob has been working as a journalist for Computerworld, covering the Danish IT industry. Originally from a small town in Jutland, Denmark, Jacob now resides in the Nørrebro neighborhood of Copenhagen. He also serves on the board of the Danish OI organization DFOI.

In his free time, Jacob enjoys dining out at Copenhagen’s many restaurants and is a fan of the Danish football club Brøndby IF.
ABBREVIATIONS

ADL = activities of daily living
BI = basilar invagination
BPI = Brief Pain Inventory Scale
BBDC = Brittle Bone Disorders Consortium
EDS = Ehler Danlos Syndrome
EFIC = European Pain Federation
EuRR-Bone = European Registries for Rare Bone and Mineral Conditions
FD = Fibrous Dysplasia
FOP = Fibrodysplasia Ossificans Progressiva
GI = gastrointestinal
HPP = hypophosphatasia
MDT = multidisciplinary
MO = multiple osteochondromas
MPQ = McGill pain questionnaire
OI = osteogenesis imperfecta
OSA = Obstructive sleep apnea
PROMIS = Patient Reported Outcomes Measurement Information System
PRO = patient reported outcome
PROMS = patient reported outcome measures
PREMS = Patient Reported Experience Measure
QOL = quality of life
SF-36 = Short Form Health Survey
STEMS = Screening Tool for Everyday Mobility and Symptoms for Skeletal Dysplasias
VAS = Visual analog scale (VAS)
XHL = X-linked hypophosphatemia
THE EUROPEAN PAIN FEDERATION’S DEFINITION OF PAIN

Pain is an unpleasant sensory and emotional experience associated with, or resembling that associated with, actual or potential tissue damage. Pain is always a personal experience that is influenced to varying degrees by biological, psychological and social factors.

Nociception refers to the nervous system’s encoding of potentially damaging events (e.g. touching a hot stove, accidentally cutting yourself). But one can experience pain without nociception and have nociception without pain. Pain does not equal nociception. Nociception is objective, but pain is subjective and does not emerge solely from activity in sensory neurons.

Through their life experiences, individuals learn the concept of pain. A person’s report of an experience as pain should be respected.

Although pain usually serves an adaptive role, it may have adverse effects on function and on social and psychological well-being. Verbal description is only one of several behaviours to express pain, as the inability to communicate does not negate the possibility that a human or a nonhuman animal experiences pain.

PAIN SCALES

Numeric pain rating scale
A person rates their pain on a scale of 0 to 10 or 0 to 5. Zero means “no pain,” and 5 or 10 means “the worst possible pain.”

Verbal Pain Intensity scale
These pain scales give people a simple way to rate their pain intensity using a verbal or visual descriptor of their pain. Some examples would be the words “mild,” “discomforting,” “distressing,” “horrible,” and “excruciating.”

Categorical scale
For children, pain scales using images of faces are commonly used. A child may be presented with the images of eight different faces with various expressions. The child chooses the face that they feel is most consistent with their current pain level.
Multidimensional tools
Some examples of multidimensional tools to measure pain include:

Initial pain assessment tool
This tool is designed for use during an initial evaluation. It helps a doctor get information from the person about the characteristics of their pain, the way the person expresses their pain, and how the pain is affecting the person’s everyday life. This pain scale includes the use of a paper diagram. It shows a body where people can mark the location of their pain, as well as a scale to rate pain intensity and a space for more comments.

Brief pain inventory (BPI)
This tool is very fast and simple for people to use to help measure pain intensity and associated disability. It includes a series of questions addressing aspects of pain felt over the previous 24 hours. See an example of this tool here.

McGill pain questionnaire (MPQ)
This is one of the most widely used multidimensional pain scales. It appears in questionnaire form, and assesses a person’s pain based on the words they use to describe their pain.

PROMIS
A set of person-centered measures that evaluates and monitors physical, mental, and social health in adults and children. It can be used with the general population and with individuals living with chronic conditions. PROMIS includes over 300 measures of physical, mental, and social health for use with the general population and with individuals living with chronic conditions. More information here.

EXPLANATIONS

Advanced therapy
Refers to new medical products that use gene therapy, cell therapy, and tissue engineering. Stem cells are defined as advanced therapy medicinal products (ATMPs).

Antibody
An antibody is a protein component of the immune system that circulates in the blood, recognizes foreign substances like bacteria and viruses, and neutralizes them. After exposure to a foreign substance, called an antigen, antibodies continue to circulate in the blood, providing protection against future exposures to that antigen.
Assessment of pain
Is a broad concept involving clinical judgement based on observation of the type, significance and context of the individual’s pain experience.

Biopsychosocial
The biopsychosocial approach systematically considers biological, psychological, and social factors and their complex interactions in understanding health, illness, and health care delivery.

Collagen
Collagen is the most abundant protein in the body. Its fiber-like structure is used to make connective tissue. Like the name implies, this type of tissue connects other tissues and is a major component of bone, skin, muscles, tendons, and cartilage.

Intramedullary canal sclerosis
Increased bone formation within the medullary cavity (the hollow part of bone that contains bone marrow)

Endpoints
The primary endpoint of a clinical trial is the endpoint for which the trial is powered. Secondary endpoints are additional endpoints, preferably also pre-specified, for which the trial may not be powered. Surrogate endpoints are trial endpoints that have outcomes that substitute for a clinical endpoint, often because studying the clinical endpoint is difficult, for example using an increase in blood pressure as a surrogate for death by cardiovascular disease, where strong evidence of a causal link exists.

EQ-5D
A standardised measure of health-related quality of life.

Measurement of pain
Approaches to the measurement of pain include verbal and numeric self-rating scales, behavioral observation scales, and physiologic responses.

Key4OI
Is a standard set of outcome measures. See more information here.

Nociceptive & neuropathic pain
The two main types of physical pain. Nociceptive pain is caused by potentially harmful stimuli being detected by nociceptors around the body. The other main type is called neuropathic. People with neuropathic pain may experience shooting, burning pain.
The pain may be constant or occur intermittently. A feeling of tingling, numbness, or a loss of sensation is also common. Neuropathic pain is usually caused by a chronic, progressive nerve disease, although it can also occur as the result of injury or infection.

**Nociplastic pain**
A long term complex pain, defined as “pain that arises from altered nociception despite no clear evidence of actual or threatened tissue damage causing the activation of peripheral nociceptors or evidence for disease or lesion of the somatosensory system causing the pain”.

**Ollier’s Disease**
Ollier’s disease manifests as greater than normal growth of the cartilage in the long bones of the legs and arms so that growth is abnormal and the outer layer (cortical bone) of the bone becomes thin and more fragile.

**Outcome measure**
An outcome measure, endpoint, effect measure or measure of effect is a measure within medical practice or research, (primarily clinical trials) which is used to assess the effect, both positive and negative, of an intervention or treatment.

**Pharmacological**
A pharmacological drug is a chemical substance used in the treatment, cure, prevention, or diagnosis of disease or used to otherwise enhance physical or mental well-being.

**Patient Reported Experience Measure (PREM)**
A measure of a patient’s perception of their personal experience of the healthcare they have received.

**Real world evidence**
Real-world evidence (RWE) in medicine is the clinical evidence regarding the usage and potential benefits or risks of a medical product derived from analysis of real-world data (RWD).

**Skeletal dysplasias**
The skeletal dysplasias are a heritable group of more than 450 well delineated disorders that affect primarily bone and cartilage, but can also have significant effects on muscle, tendons and ligaments

**Transition**
The process of changing from a paediatric to an adult model of health care.
BALANCING LIFE WITH OI

Topical meeting on the impact of pain in osteogenesis imperfecta (OI)

9-10 June 2023 - Stockholm