Editorial

By Jacob Wittorff, OIFE Board Member

IS IT TIME FOR A EUROPEANS WITH DISABILITIES ACT?

For years, I just took the world as it was. When faced with a flight of stairs or other obstacles, I simply shrugged and turned around. But at some point, I realized it doesn’t have to be this way. The world can look different. One of the things that helped me recognize this, was traveling in the USA.

In Denmark, where I live, it often feels like we put the welfare state on a pedestal. Lots of Danes find it hard to believe that there are places where people with disabilities or other challenges, have better opportunities to participate in society than here in our Nordic welfare states. Therefore, Danes are often surprised when I tell them that accessibility for wheelchair users is often much better in the USA than here in the very Nordic welfare states, which progressive American politicians like to highlight as a role model.

Part of the reason for the high accessibility is The Americans with Disabilities Act (ADA), passed in 1990. Among many other things, it mandates that all public buildings must be accessible to people with disabilities. As shown in the excellent American documentary Crip Camp, this legislation is the result of a years-long rights struggle.
Traveling in the USA has always been a pleasure for me because all hotels have accessible rooms (albeit often of varying quality). When I encounter a staircase in the USA, I automatically look for the elevator or the ramp. In Denmark, I often prepare to turn around and go home. And that has happened quite often. A few years ago, a survey showed that over half of the shops on the main shopping street in Copenhagen are inaccessible for people in wheelchairs.

Let me stress: This is not meant to be a lament. I’m immensely grateful to have been born in Europe, and perhaps especially in the Nordics, where among many other things, I have access to free healthcare and receive modern mobility aids paid for by the public.

The Icelanding entrepreneur Haraldur Thorleifsson, who was born with muscular dystrophy and sold his company to Twitter a few years ago, has taken matters into his own hands. He started his own project, Ramp Up Iceland, with the original ambition to fund the construction of 1000 ramps in Iceland. Recently, he expanded the project. With help from the NGO Start Small, he is now trying to spread his project to the rest of Europe, initially to three yet unnamed major European cities.

Haraldur Thorleifsson’s project is commendable. However, it also raises the question of whether it’s time for Europe to emulate the USA by assuming political leadership and regulating this area.

A small step was taken in 2019 when the European Accessibility Act was passed, focusing mainly on making digital products and services user-friendly for people with disabilities.

But isn’t it about time we introduce a European With Disabilities Act, something on par with the American ADA?

A piece of legislation ensuring that we, as EU citizens with disabilities, can move and travel freely, whether we live in Helsinki in the north or Lisbon in the south.

Next summer, as we face an election to the EU Parliament, there is hardly a better time to ask our candidates what they intend to do to improve accessibility for EU citizens with disabilities.

Jacob Wittorff
OIFE Board Member

What is the OIFE doing?
By Ingunn Westerheim, OIFE President

Between September and November, things have been a little bit less busy trying to catch up with various research projects and post conference activities after the Stockholm conference. On August 25th we had the pleasure to attend a business lunch and 12th the anniversary of the Spanish foundation Fundacion Ahuce in their offices in Valencia. After that we have continued to work on the IMPACT project and the Pain Project. We have also been busy planning and hosting events like the OIFE Clinical Trial Update and the planning of 2nd OIFE Investigator Meeting.

At the end of October we returned to Spain for the World Orphan Drugs Congress Europe, where OIFE was represented by Ingunn Westerheim, Stephanie Claeys and Inger-Margrethe S. Paulsen.
And from November 24-26 five of the board members finally had the chance to meet face to face again at our Board meeting in Mechelen, Belgium. We discussed strategies and plans for next year, which include an online AGM (part one) and a face to face meeting (part 2) in beautiful Valencia.

Meetings we have organized or attended between September and mid-November:

- OIFE Board Meetings (Zoom), Sep 19, Oct 17 and November 25-26 (face to face)
- Planning OIFE Investigator meetings Sep 13th + Oct 24 (IM, JC and IMSP)
- NFOI Adult Meeting, Sep 16th (IW attended face to face and gave a talk about OIFE)
- VC Global Pediatric Endocrinology and Diabetes (GPED), Aug 18 (IW)
- VC HR-pQCT Working Group, October 5 (IW and RTS)
- RBD Echo New Therapies, October 5 (IW)
- VC Isala clinic & VOI, October 6 (IW and TvW)
- VC European Society of Endocrinology (ESE), Oct 12 (IW)
- VC European Federation of Pharmaceutical Industries & Associations (EFPIA), Oct 13 (IW)
- Ultragenyx press conference ORBIT, Oct 16 (IW and TvW)
- VC Boost Pharma, Oct 17 and Nov 14 (IW)
- OIF hybrid conference about transition, Oct 19 (IW attended 1 session)
- VC IMPACT survey, steering committee Oct 25 (IW and TvW)
- VC Rare Revolution Magazine, Nov 9 (IW)

WHAT DOES THE OIFE MEAN TO YOU?

What does the OIFE mean to you? This is the question Ingunn asked in the editorial of the previous OIFE Magazine. It is also the main question in the new video about OIFE.

Now you can find the brand new video "What is OIFE?" on our website. Perhaps you will find some familiar faces there?
WORLD ORPHAN DRUGS CONGRESS IN BARCELONA

From October 30\textsuperscript{th} to November 2\textsuperscript{nd} we returned to Spain for the World Orphan Drugs Congress (WODC) Europe, where OIFE was represented by Ingunn, Stephanie and Inger-Margrethe. WODC is primarily for the industry, there are also policy makers, NGOs and other types of stakeholders present. It is a huge conference with more than 2000 participants and this year it took place in a massive hall with 10 “tents for” parallel sessions, which brought some logistical challenges including bad sound. A nightmare for those who were hearing impaired. But if you disregard the lack of food and coffee (we had free tickets as patient representatives anyway), the conference was both interesting and thought provoking with excellent opportunities to meet new and old contacts.

The event started with a smaller workshop about how better use of data can reduce the impact of rare conditions. And during Q&A OIFE shared our experiences from the IMPACT survey, which created a lot of interest among the participants and brought us new contacts.

Day 2, 3 and 4 were packed with plenary lectures, parallel sessions and networking opportunities both on site, but also at the reception at the National art museum of Barcelona. Topics included the new European pharmaceutical regulation, advanced therapies, the use of real world evidence, access to new therapies, and patient involvement in clinical trials, in health technology assessment (HTA) and in development of policies and regulations. The last day, we attended an interesting panel debate about adult care and services (or lack of such) for rare conditions. It also touched upon the complexities of getting older with a rare condition. And the need for more and better data and natural history studies.
OIFE CLINICAL TRIAL UPDATE
On October 24 we managed to gather 125 people from a large number of countries for our OIFE Webinar about Clinical Trials. The goal of the webinar was to update the OI-community about the following clinical trials and projects:
- The Cosmic and the Orbit trials (setrusumab)
- The BoostB4 trial (stem cells)
- The Topaz trial (teriparatide and zolendronate)
- The Poise 1 study (SAR439459)
- The Saturn project (real world evidence)

You can read a report from the webinar another place in the magazine.

OIFE INVESTIGATOR MEETING
We feel it is fair to say that also this year’s edition of the OIFE Investigator Meeting was a success. We had more than 160 people attending, from a huge number of countries. Prominent researchers from Europe, USA and Asia, shared their knowledge about basic and clinical research. And the meeting ended with a constructive panel debate about how we can develop a common classification system for OI types that both researchers, clinicians and people with OI and their families are happy with. Thank you to the committee, the speakers, the moderators, the volunteers and the participants for making this an interesting and interactive meeting! Read more about the event in the next OIFE magazine.

OIFE AT EFPIA PATIENT THINK TANK
November 22nd we had the great pleasure of being invited as observers to the hybrid meeting of the Patient Think Tank of European Federation of Pharmaceutical Industries and Associations (EFPIA). We learned about the work of other European federations, hurdles and opportunities for how patient organizations can work with industry, medicine shortages and patient engagement in health technology assessments (HTA) on a European and national basis.

EFPIA PATIENT THINK TANK
A forum for a co-creation, and an open exchange of ideas, information and perspectives between Patient Organisations and Industry on topical issues impact on patients

MISSION:
To drive best practice in working together where the patient is integral to decision-making. This extends to EFPIA policy development across the life-cycle of a medicine, through development, access and use.

VISION:
Better health outcomes driven by meaningful, balanced and equal cooperation between industry and patient organisations.

VALUES:
- Clarity of purpose
- Transparency
- Independence
- Respect
- Accountability
- Inclusiveness
- Continuity, commitment and consistency
BILBAO CONFERENCE ON THE FUTURE OF EUROPEAN REFERENCE NETWORKS

On October 11th EU leaders and key policymakers gathered in Bilbao, Spain, for the Conference on Rare Diseases and the European Reference Networks taking place under the Spanish EU Council Presidency. Discussions addressed some of the main challenges facing the EU policy framework on rare diseases, explored the future of the European Reference Networks (ERNs), and answered calls for action from the previous Czech Presidency of the EU Council. Representatives from OIFE attended online.

In anticipation of the conference EURORDIS and the 24 ERNs wrote an open letter to EU institutions about why they must “stand by” the ERNs and renew their support for them over the coming years. OIFE completely supports the message from EURORDIS.

ANOI (OI BRAZIL) NEW OIFE MEMBER

We are very happy to announce that the national OI-organization of Brazil Associação Nacional de Osteogênese Imperfeita - ANOI has been approved as a new associate member organization of OIFE. ANOI has been formally registered since 2017 and has more than 1100 members with OI! A very warm welcome to our growing international OI community!

OIFE IN VALENCIA

On August 25th we were happy to be invited to the 12th anniversary of the Spanish foundation Fundación Ahuce, which took place in their offices in Valencia, Spain. Before the celebration we were invited to a business lunch with representatives from Ahuce Foundation, dr. Ana Bueno, OIFE volunteer Maria Barbero and representatives from Mereo Biopharma.

It was our pleasure to be there and celebrate with you together with new and old friends! And we will never forget the world’s largest paella.
Enpr-EMA Annual Meeting on Pediatric Research
By Miguel R. Molina, member of OIFE’s Medical Advisory Board

October 10th Miguel R. Molina from our Medical Advisory Board represented OIFE at the meeting of the European Network of Paediatric Research at the European Medicines Agency (Enpr-EMA) in Amsterdam.

The European Network of Paediatric Research at the European Medicines Agency (Enpr-EMA) is a network of research networks, investigators and centres with recognised expertise in performing clinical studies in children. Enpr-EMA enables networking and collaboration with members from within and outside the European Union (EU), including academia and the pharmaceutical industry. It acts as a platform for sharing good practices as well as a pan-European voice for promoting research into medicines for children. At this present moment there are 21 Networks working in the Enpr-EMA with 15 European countries represented in national networks also including Japan, Switzerland and the United States. Here are some of the take-away messages from Miguel:

OFF-LABEL EVIDENCE
Off-label is not always off-evidence. Half of all paediatric medicine prescriptions in the EU are off-label. Today legislation mandates clinical trials for those medicines that are still under patent protection. The proposal of this working group is trying to include more paediatric data. Steps from off-label to effective and safe therapies can be:
- Step 1: A decision model for evidence generation.
- Step 2: How to prioritize international consensus?
- Step 3: Research agenda by subspeciality
- Step 4: Generate the evidence
- Step 5: Implement in practice
- Step 6: Market authorization

OTHER WORKING GROUPS
Other topics covered by the Enpr-EMA working groups:
- Survey of paediatric clinical trial site requirements
- Survey done on EU Research Nurses
- Paediatric clinical trial sites incl. workshop co-organised with Conect4children (C4C)
- Language discrimination and cross-border access to paediatric clinical trials

DECENTRALISED TRIALS
Another topic was the decentralised elements in clinical trials a paediatric perspective. We need to take advantage of technological progress to make clinical trials more accessible and participation more convenient for trial participants. This includes trials performed more regionally or locally, more use of wearables, home nursing, patient reported outcomes, new forms of digital consent, administration of medicinal products at home etc.
Decentralisation has huge potential for minimizing logistical hurdles affecting families. Indirect costs are significantly higher for families with children enrolled in a trial compared to adults. Because “A sick child is a sick family.”

REGULATORY AFFAIRS
This second part of the session was about the Good Clinical Practice – Scientific Guideline ICH E6 and the Accelerating Clinical Trials Project. Then there was a talk and discussion about the new EU pharmaceutical legislation and its impact on paediatric and orphan medicine development. The consultation about the new regulation will probably be in the EU parliament in 2025. And probably the new legislation could be implemented in 2027, but this is not for certain.

CONNECT4CHILDREN
The third session was about highlights in 2022-2023 of Enpr-EMA and its member networks incl. the Conect4Children journey. The C4C academy has shown to be very useful for students and researchers. You can find more information about this here.

PATIENT INVOLVEMENT
Session four was about patient involvement which included the following topics:

- Patient recruitment and retention.
- Benefits of involving patients and young people (YPAGs) in paediatric clinical research. Through the EYPAGNET 14 countries and more than 30 groups are working with patient involvement of young people.
- Improvement of paediatric health, medicine research, and innovation by sharing children’s voices
OIFE’s Clinical Trials update webinar – a summary
By Gabriela Beug

On the 24th of October, the OIFE hosted a webinar for the OI community about clinical trials and updates. 235 people from 52 countries signed up and ca 125 attended. Attendees were a mix of health professionals, scientists, OI clinicians, industry professionals, and individuals with OI and their family members.

OIFE is excited to share this current OI study and research update with you. Some of the studies are currently recruiting, see announcements another place in the magazine.

The Orbit & Cosmic trials – Ultragenyx & Mereo

Ultragenyx is in collaboration with Mereo Biopharma, conducting two clinical studies, ORBIT and COSMIC, to investigate setrusumab (UX143), a monoclonal antibody, for the treatment of osteogenesis imperfecta (OI) in pediatric and young adults.

The objective of both ORBIT and COSMIC is to understand how setrusumab can safely and effectively restore bone production and reduce fracture risk. Setrusumab is hypothesized to block sclerostin, a protein that slows the process of bone forming. It is thought that blocking sclerostin will allow increased bone formation and stronger bones, which in turn might reduce fracture risk.

Ultragenyx is responsible for planning and implementing the clinical trial globally. They also own the legal rights to the compound globally, except in Europe, where Mereo still owns the rights.

ORBIT Study
Phase II/III study
The purpose of the ORBIT study is to assess dose, efficacy and safety of setrusumab vs. placebo.

- This study is currently enrolling patients aged 5 to <26 years
- Individuals must have a confirmed genetic (COL1A1 or COL1A2) diagnosis of Type I, III or IV OI
- Must be willing to not receive bisphosphonate therapy during the study*
*Individuals are expected to stop bisphosphonate use during this study, so that solely the effect of setrumubab can be analyzed. The use of multiple medications that impact bones make it difficult to interpret results.*

Learn more about the study [here](#).

**Ultragenyx has announced promising interim data from Phase II Orbit study**

In their press release on October 14, 2023, Ultragenyx announced that treatment with setrumubab reduced incidence of fractures in patients with OI by 67% following at least 6 months of treatment and demonstrated improvements in lumbar spine bone mineral density (BMD).

**COSMIC Study**

**Phase III study**

The purpose of the COSMIC study is to evaluate the efficacy of setrumubab versus intravenous bisphosphonates (IV-BP) in children with OI.

- This study is currently enrolling patients aged 2 to <5 years
- Individuals must have a confirmed genetic (COL1A1 or COL1A2) diagnosis of Type I, III or IV OI
- Prior/current exposure to bisphosphonate therapy

Learn more about this study [here](#).

To learn more about Ultragenyx research, visit [https://www.ultraclinicaltrials.com/OI](https://www.ultraclinicaltrials.com/OI) or reach out via email, [OIStudyInfo@ultragenyx.com](mailto:OIStudyInfo@ultragenyx.com).

**Poise 1 (Sanofi)**

*TGF-β Inhibition for the Treatment of Osteogenesis Imperfecta*

Sanofi is conducting an early phase study in adults with OI Types I and IV with an anti-TGFβ antibody called SAR439459. This study is called **Poise 1** and is a Phase 1 study, where the researchers evaluate the treatment’s safety and determine a safe dosage range.

**Inclusion criteria:**

- Male or female between 18 and 65 years old, with the exception of post-menopausal women
- Confirmed diagnosis of OI Types I or IV, including documented genetic mutation in the COL1A1 or COL1A2 genes. Sanofi will provide OI-specific genotyping if it is not already in the patient’s medical history.
- Have experienced ≥ 2 bone fractures since the age of 18 OR at least 1 bone fraction in the last 10 years.

This study involves a single administration of SAR439459 given intravenously (IV) into the arm, with a 6-month follow period. At this early stage in development, Sanofi is recruiting a limited range of study participants, but they will consider expanding enrolment criteria in future studies.

Participants in the Poise 1 study are not likely to experience benefits from SAR439459, and 25% will receive a placebo, but all participants will help with the scientific understanding of OI and SAR439459 as we prepare for future long-term studies. The assessments in this study include digital, non-invasive strategies to better understand how OI patients move and are active throughout the day as well as direct patient feedback on daily activity and pain.
TGFβ is a signalling molecule, which is a way cells communicate and coordinate with each other. Specifically, it is an important part of the bone remodelling environment, playing a role in the balance of forces which remove and build new bone. It even has a role in pain. In OI, signalling related to TGFβ is dysregulated, so controlling that signalling with SAR439459 may be a way to influence symptoms caused by OI.

Study participants will be compensated for their travel and accommodation associated with visits to the study site. Such travel and accommodation can be arranged directly by Sanofi or a third-party service provider appointed by Sanofi.

For this early study, our two sites in Europe are located in France. The other sites are in the US, Canada and Australia. Additional information on participating study sites and how to contact Sanofi, if interested, is available here.

**The TOPaZ Trial**

*Treatment of Osteogenesis Imperfecta with Parathyroid hormone and Zoledronic acid*

TOPaZ is the largest study performed so far in osteogenesis imperfecta with 350 participants. The purpose of the TOPaZ Trial is to investigate whether a two-year course of teriparatide (TPTD) followed by antiresorptive therapy with a single infusion of zoledronic acid (ZA) in adults with OI reduces the risk of fracture as compared with standard care. Teriparatide is effective at increasing bone mineral density (BMD) in OI and Zoledronic acid is hypothesized to be able maintain a new bone after it has been formed. Almost anyone >18 with a clinical diagnosis of OI is eligible. See more info on the project webpage.

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**BOOSTB4 Update**

Boost Brittle Bones Before Birth (BOOSTB4) is a research study investigating the possibility of using highly bone forming fetal mesenchymal stem cells (fMSC) as a treatment of severe OI.
The treatment involves transplanting the stem cells into the baby affected with OI during pregnancy or soon after birth. The main aim of the BOOSTB4 clinical trial is to test whether the treatment is safe and effective.

The BOOSTB4 study has completed participant recruitment. In the primary follow-up of patients, no serious or adverse events were observed, the treatment was deemed safe after multiple i.v. booster doses, no immune response to the mismatched fetal-MSC occurred and only minimal pain was reported. See more information at the project webpage or at clinicaltrials.gov.

**Project Saturn**
Project Saturn is a real-world evidence and data collaboration with existing European datasets of osteogenesis imperfecta to support future therapies and answer specific research questions.

For the timely and sustainable availability of therapies, the agreement of stakeholders is necessary. These stakeholders are Regulators (ETA), Health Technology Assessments (HTAs), and Payers. The size of clinical studies for rare diseases is usually limited (+/- 200 patients). Health Technology Assessors (HTAs) and Payers would likely need to see more evidence/data before granting access and making therapies available long-term.

The objective of Project Saturn is to meet the data needs of all stakeholders, to ensure they have the evidence to support the decision to make a potential therapy available to patients and to converse with data stakeholders to understand what data is needed. This in turn, will make new therapies available to the people who need them. Find out more by contacting Mereo Biopharma.

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**Balancing life with OI – now on YouTube**

The 2023 OIFE’s Topical Meeting “Balancing life with OI” was the first conference of its kind to cover the impact of pain in OI. Most of the talks and presentations are now available with English subtitles in OIFE’s Playlist on YouTube. The various talks cover the causes, assessment, and management of pain in OI and other rare bone conditions. This in addition to pain’s impact on physical and mental health, sleep, fatigue, mobility, relationships/families, and work/life balance.
For your convenience, below you find the categorized links to the talks. You can have auto texting in other languages, by clicking on ‘settings’ in YouTube:

**Pain: introduction to pain in OI and other rare bone conditions**

- What is pain - Audun Stubhaug
- What are the causes and different types of pain in OI - Jacqui Clinch
- A multicenter study to evaluate pain characteristics in OI - Mercedes Rodriguez Celin
- Chronic pain in adults with OI - appraisal, coping, and QoL - Ruben Munoz Cortes

**Assessment and measurement of acute and chronic pain**

- How to assess pain: pros and cons of different tools - Mercedes Rodriguez Celin
- The assessment of pain in children with osteogenesis imperfecta - Kelly Thorstad
- Assessing pain in adults with osteogenesis imperfecta - Richard Keen
- Sleep quality, daytime sleepiness and increased risk for OSA in patients with OI - Linda Lušić
- Evaluation of mobility associated symptoms in Argentinian OI-patients - Mercedes Rodriguez Celin

**More than just bone pain**

- Neuromuscular health and function in children with osteogenesis imperfecta - Alex Ireland
- Pain and basilar invagination in osteogenesis imperfecta - Eva Åström
- Osteogenesis imperfecta and gastrointestinal pain – what do we (not) know - Lena Lande Wekre
- Perceived dental care needs and concerns of individuals with OI - Coreen Kelday
- Intramedullary canal sclerosis - a complication of prolonged bisphosphonate therapy - Željko Jeleč

**The IMPACT of pain in OI**

- IMPACT Survey background and overview - Samantha Prince
- Connection between pain, mental health, sleep and fatigue - Andrew Wiese
- Balancing ambitions with pain in OI - Karen Braitmayer & Jacob Wittorff
- Parenting a child in pain - Kis Holm Laursen
- Pain’s impact on relationships and sexuality - AnnBett Kirkebæk
- The Pain and OI Survey - Michael Stewart
Managing pain in OI

- How we work in the MDT pain clinic Bristol and Oslo - Clinch and Stubhaug
- Modalities to deal with pain caused by fractures and surgeries - Cathy Raggio
- Pharmacological pain management - Richard Keen
- Therapeutic pain management principles in osteogenesis imperfecta - Sophie Barlow
- Physical activity - A good natural medicine for pain and health in OI - Miguel Rodrigues Molina
- Educational resources for children with OI experiencing acute and chronic pain - Kelly Thorstad
- A toolbox for pain in OI empowering patients to take charge - Ariane Kwiet
- Transition and Follow up in Adult OI - Jannie Hald
- Impact of advanced therapy on pain in OI - Clara Rodríguez

Other topics

- Similarities and differences between OI and other bone conditions - Alison Boyce
- Use of medical care by individuals with OI - Silvia Storoni

The abstract book of the meeting with summaries from all the speakers can be downloaded for free here. More on this topic can be found in OIFE’s The Pain & OI Project
Meet the OIFE Delegates – Associate Member Panama

JESUS CAMILO OSORIO BARAHONA

Tell us a bit about yourself!
I am Jesús Camilo Osorio Barahona, I am a happy husband, being married to Daysi, a woman that supports me and believes in me. I am the father of two beautiful daughters, Lluvia and Luna (one of which has OI) and I have OI myself. I've been lucky to have my parents, my wife and my family around me and they are my reason to work hard every day. I am a creative person with a vision to improve the quality of life of the people with OI in Panama.

Tell us a little about the organization you represent!
OI Panama works as an organization for people with OI, providing them with emotional support, guidance, and updated information on the latest scientific advances in relation to OI. Furthermore, it serves as an awareness platform in schools and companies to promote inclusion at the national level in education and the work environment.

Are there issues people with OI must struggle with in Panama? Do they get support?
People in rural areas have difficulties finding treatment and therapy for their families and the lack of awareness in schools make their situation very difficult. But they do get some support from the government and institutions like the National Rehabilitation and Physical Medicine Institute where they can get therapies at low cost and support from a specialist who understands the condition. Also, the Panamanian Social Security Fund provides treatment with bisphosphonates to improve the life quality of the patients.

What do you do when you're not working for OI Panama?
I am a full-time chess instructor and player. Chess has changed my life. It has given me the opportunity to test myself and travel to different places with my talent and inspire other people through my Social Networks like Tiktok, where I have almost 39,000 followers.

In your opinion, what is the most important task for national OI organizations?
To Promote the creation of public policy and create alliances with private entities to create projects for the benefit of our population.
Above is a poster featuring Luna (daughter of Jesus), in her national costume, who is part of the new campaign of Ayudas Panama. They are the Rare Disease Umbrella of Panama, and the campaign is focusing on inclusion and diversity of different ethnicities, cultures and disabilities.

We congratulate the new OI organization in Panama Familias OI Panamá with their successful inauguration meeting on October 21st. Many people were present and even more followed online. OIFE also contributed with a small video greeting, wishing the new Board good luck with creating a strong OI-community in Panama.

**Adult Health resource group**

*Interview with Eric Orwoll, Professor of Medicine, Division of Endocrinology, Diabetes and Clinical Nutrition, School of Medicine and Dr. Lars Folkestad, member of OIFE’s Medical Advisory Board*

Due to common interests in adults with OI, Prof. Eric Orwoll and Dr. Lars Folkestad identified researchers and clinicians who have worked on adult care and formed a resource group. During the summer of 2022 the group started meeting online to discuss a potential collaboration to identify the gaps in knowledge related to adult care.

**Tell about this initiative to uncover knowledge gaps in research about adults with OI!**

**Who is in the group and how do you work?**

Due to common interests in adults with OI, we identified researchers and clinicians who have worked on adult care. During the summer of 2022 the group started meeting online to discuss a potential collaboration to identify the gaps in knowledge related to adult care. The aim of the group is to establish a cross Atlantic collaboration between different groups.

The group consists of the Chair Eric Orwoll and members from the US, Laura Tosi, Cathleen Raggio, Sandesh Nagamani and from Europe, Lars Folkestad, Jannie Dahl Hald, Bente Langdahl, Stuart Ralston and Oliver Semler. The OI Foundation (OIF) has been instrumental in facilitating the meetings. They are now held on a regular basis, and several projects are ongoing. The aim here is to become wiser together and share knowledge and experiences to strengthen our research.
What is the mandate and goals?
We do not have an official mandate, but the group members share a common goal – to increase the quality of care for adults with OI. This is a research collaboration. We aim to produce high level publications that will inform decision support for clinicians and patients alike, and to identify priority areas of research to advance knowledge of OI in adults.

You published a position paper called ‘Osteogenesis Imperfecta Is More Than a Pediatric Disorder—There Is a Need for Adult Care Evidence to Guide Clinicians’ - tell us about it!

Our commentary was meant to highlight some of the gaps in knowledge related to adults with OI. We find that ageing adults with OI have unique challenges related to having OI, and that these challenges may differ between different age groups. This needs to be further evaluated. Our commentary highlights a few of these issues and we hope to get funding agencies to share our interest in ensuring that future research is focused on these issues.

We often say that OI is 2 very different conditions for children and adults - do you agree?
As stated above, we agree with OIFE that different ages have their own challenges. For instance, in children with OI support of growth is essential. On the other hand, adults face challenges that are associated with aging and frailty. In the presence of conditions that are associated OI, ageing may influence adults with OI differently than the non-OI population. Take osteoarthritis for instance, we know that there is a link between risk of osteoarthritis and age – the older you get the more osteoarthritic you become. In OI the collagen defect in itself may influence the risk of osteoarthritis and signs of the condition may come at an earlier age. The next question then, is how we then prevent and treat these conditions in adults with OI.

Data from the IMPACT survey

The regulators EMA and FDA emphasize counting fractures to prove that new treatments are efficient - what are the challenges around this, especially when it comes to adults?
That is a difficult question. New treatments need to be effective, thus fracture preventing drugs should prevent fractures, right? Studies designed to show a reduction in fracture rates often need to be large, something that is difficult in a rare disease.
Furthermore, epidemiological studies of OI have shown that fracture rates go down after adolescence, making it especially difficult in adults to conduct sufficiently large studies to identify a drug’s effect on fracture rates. Even so, fractures are more common in adults with OI than in unaffected people, so studies intended to demonstrate fracture risk reduction are critical.

**When and how should BPs in adults be considered - X-ray, DXA, biomarkers or other ways?**
That is another great question, with no 100% correct answer. The decision to initiate treatment is always the result of an individual assessment of the patient based on medical history, imaging, fracture history, family history, and other risk factors. However, while we know a lot about the association between common risk factors, bone density and fracture risk in age related osteoporosis – there is little evidence related to these issues in the OI field. Nevertheless, we try to use the same principles to identify those with OI who are at high fracture risk and consider them for bisphosphonate (or other) therapy. Clearly, this problem needs to be studied in more detail.

**There are several initiatives (ERN BOND, Care4BB etc.) working on developing guidelines and recommendations for adults with OI.**

**Is this even possible to develop official guidelines with the current lack of research?**
It is indeed very difficult to confidently develop guidelines without a strong foundation of good research. This is one of the problems that motivated the formation of our Adult OI group. We want to carefully evaluate major areas of concern in OI and to identify critical research needs. Still, even without a complete foundation in research, there is a real need for clinical guidance. So, in addition to identifying research directions, we also hope to use whatever data are available to provide suggestions to help patients and clinicians to manage problems facing adults with OI.

**Different countries have different definitions of guidelines - can you explain?**
Guidelines are exactly that – guides to ensure the highest quality of care. However, there may be national factors such as number of specialists, scanners, laboratories etc. that may influence what choices clinicians can make related to patient care.

**Are recommendations based on best practice more realistic?**
Lack of evidence is an important limitation to what the guideline can incorporate, and here best practice or consensus between experts is often the best option in developing guidelines. But again, our body of evidence is based on research on a group or a population. And treatment/follow-up choices for the individual patient are made based on the clinician’s experience, knowledge, and in collaboration with the person seeking help, trying to translate what we know from the evidence into what would be the best option for the individual patient.

**What can we do while waiting for the uncovering of knowledge gaps and the following much needed research?**
That question makes it sound like we are roaming around in the dark, and that is not true. There is a lot of knowledge available, and new studies are initiated and published almost every day. Our working group is trying to find ways to highlight the knowledge that is available as well as to identify areas where additional studies could help chose between treatments and follow-up regimes with more certainty to improve the quality of life for adults with OI.

**Besides those mentioned. What are the main knowledge gaps regarding adults with OI?**
Sometimes when you shine your light out in the dark you find things you did not know were there. We all need to listen to the stories that we are told by the adults with OI.
We know that the patient community will keep the clinicians and researchers on their toes to keep the research relevant for the patient community.

**How can we as a patient community - help in the most effective way?**

OIFE, OIF and C4BB are doing a great job by facilitating meetings globally. The organizations’ interest in research and in expanding expertise is invaluable for the scientific community. Participation in the scientific meetings, the discussion at these meetings and the willingness to participate in research project planning, organization, running and knowledge dissemination is much appreciated.

![Assessment of Pain](image1)

**Voice and OI**  
*Interview with Katharina Böhm*

**Who are you & what is your relationship to OI?**  
My name is Katharina Böhm, I am 25 years old and have studied speech therapy. Since I have OI type 1 myself, I have always had a connection to the subject.

**Tell us about your project!**  
I wrote this thesis as part of my bachelor's degree in speech therapy at the OTH Regensburg in Bavaria, Germany. I did the research on my own and my examiner Prof. Dr. Norina Lauer was available to help me with any queries.

Most of the research took place within the framework of two events of the German Society for OI (DOIG). In the quantitative study, the voice function of people with OI was recorded on the basis of two voice recordings with the computer program Praat. The Acoustic Voice Quality Index (AVQI; version 03.01) was calculated and numerous other parameters such as pitch, loudness and exhalation duration were examined. Socio-demographic and health data as well as personal experiences with and satisfaction with one’s own voice were recorded by means of short questionnaires.

I initially financed the trips myself and was graciously given a large grant at the end to cover the costs of traveling through Germany. I was also able to borrow a microphone for the voice recordings from the OTH Regensburg.
What was your research project about?
Due to the phenomenon of the "higher pitched voice" described in the literature, the question arose in my mind whether the voice is altered in OI. The aim of this study was to find out by means of voice analyses whether there are pathological changes in the voice sound in people with OI. In addition, the subjective assessment of the test persons was to determine whether and why there might be suffering pressure in order to uncover a possible need for action in the field of speech therapy and to enable better care. The target group were all adults with OI from Germany.

What were your most interesting findings?
Forty-seven individuals with OI from Germany with a height between 80 cm and 174 cm (M = 131.36 cm, SD = 26.55) who were over 18 years of age were studied. Scoliosis was present in 85% of the respondents. 36 affected individuals (n = 47) exceeded the AVQI (v03.01) cut-off value of 1.85. There was also an increase in speaking voice pitch in males (M = 128 Hz, SD = 18.79), whereas females were within the normal range (M = 207.15 Hz, SD = 23.34). The mean speech loudness of the affected subjects was significantly elevated at 70.73 dB (SD = 3.50).

No major differences were shown between the experienced positive and negative feedback on one's own voice. Exhalation duration decreased significantly the more severe the scoliosis, the smaller the body size, and the more frequently a wheelchair was used. Men with elevated voice experienced significantly fewer positive experiences related to their voice (p = .007). Satisfaction with their own voice was in the upper third, with some outliers in the negative direction.

Any surprising findings?
I found it exciting that the personal experiences were so different. Some people suffer a lot, others are very happy with their voice.

What is the most important take home message for clinical work?
A speech therapy as well as respiratory therapy treatment could support some affected individuals with significant distress. Further research, especially a close examination of the anatomical conditions, with regards to an effective treatment might be useful.
Were patients/patient organizations involved? How?
The German Society for Osteogenesis Imperfecta (DOIG) was involved because I know a lot of people with OI through my own membership and was able to contact them. In addition, I was given the opportunity to examine all people with OI who agreed to participate in the study at two meetings. I was able to examine the OI people at these meetings and visit some of them on a tour through Germany and make voice recordings at home. For this opportunity and all who supported me, again a big thank you!

Any messages for the readers of OIFE Magazine?
Now I would be interested to know if you have also noticed this phenomenon. Are you satisfied with your voice, or would you like to see more research in this area? You are welcome to write me your thoughts via Email. I will be writing my master’s thesis in a few years, and hopefully I could continue to write and research in this area if you are interested. For this, feedback on my study would be very helpful.

OIFE Youth Event
By Malene Sillas Jensen & Lars Nesset Romundstad

Why OIFE youth events?
For close to two decades, there have been different events organised by national organisations, or in collaboration with OIFE all around Europe. Countries such as Germany, Belgium, the UK, Norway, Denmark, the Netherlands and now Italy have all hosted young people with OI. It is globally regarded as an excellent platform for young people to come together, share experiences, and to exchange ideas and see ideas from different perspectives.

For many, it is also regarded as a safe, inclusive space for people not accustomed to travelling alone or with a personal assistant. In addition, with support from OIFE and others, it has traditionally also been a space which has allowed people from all over Europe to come for a reasonable cost, compared to an equivalent trip arranged in a private capacity. An additional rationale for national organisations to send delegates to the events has also been to increase interest and knowledge in the field of OI, both internationally and nationally. In many countries, participants in these events have been, or did take up positions in boards and committees in their respective countries, in the theme of OI. In Italy, as before, this was accomplished through long discussion far into the night, new friendships and ideas spreading across borders.
Because on the 7th of September 2023 we met in Italy!
It was once again time for the young people of the European OI community to come together. This time in Lido di Jesolo, in the Veneto region of Italy, one hour away from Venice. The event was organised by the Italian OI organisation Associazione Italiana Osteogenesi Imperfetta (ASITOI), and 30 participants from 12 countries made their way to the sunny Italian coast by both plane, train and car. Similar to years before, both seasoned OI youth meeting participants, as well as new ones were excited for the next few days, having the opportunity to get to know each other, exchange experiences and learn from each other on OI-related topics, and completely different things. The countries of Italy, Switzerland, Denmark, Norway, Poland, Germany, The Netherlands, Belgium, The United Kingdom, Ireland, Croatia and Finland were all represented, and were in the age group 16-35 years old.

The meeting was held in the magnificent resort Villagio al mare Marzotto
This resort is a highly equipped compound with a football field, restaurant and bar, pools, basketball courts, and located right at a beach with astonishingly accessible facilities. We were split into different rooms, some living with pre-arranged roommates, others with people they had never met before.

Read the rest of the report from OIFE Youth Event on our website.
Other rare bone conditions than OI: FOP

Interview with Marie Fahlberg

Marie Fahlberg, President of the Swedish FOP association attended OIFE’s “Balancing Life with OI” conference in Stockholm. She says that she learned a lot about similarities between rare bone disease. For instance, getting to understand the holistic perspective of the rare bone diseases and not just focus too much on the bones.

What is your relation to a rare bone condition?
My name is Marie H. Fahlberg and I am from Sweden. My youngest son Hugo was born with Fibrodysplasia Ossificans Progressiva (FOP) in 1998 and diagnosed in 2001.

In a few words - tell us about FOP and how it's affecting lives?
FOP transforms muscles, tendons and ligaments into bone. It creates an extra skeleton and bridges of bones lock the joints. The most common sign of FOP at birth is a malformation of the big toes, but there are variations too without the FOP-toes. A person with FOP needs to be careful in many ways. They need to avoid intramedullary injections, biopsies and surgeries. These actions and even small traumas and viruses can trigger FOP to start to form new bones. The transformation is often painful with soft tissue swellings. Sometimes the swellings are small lumps, other times they are large and can cover a whole back. The swellings and bone formations can overlap and continue for months and years before it calms down for a while, and you never know for how long or when it will start again. It is different from person to person.

As a parent to a small child, it’s a lot about creating a safe environment, to have a person look after the child all time, and informing about FOP to avoid unnecessary accidents, swellings and bone formations. But FOP is unpredictable; it can “wake up” and start a flare up one day without any warning.
Many of the adults are very stiff. Some still have movements in the hips and are able to walk. Some are stiff in a seated or standing position and in need of a wheelchair or Permobil (powerchair). All of them need an assisting person to help manage their daily life on many levels.

But it is important to stress that FOP is only physical; it does not affect the brain. And the International FOP community is amazing in many ways. It is like an extra family.

**How rare is FOP?**
Approximately 1 in 1 million.

**Which organizations are you involved in?**
I am the President of the Swedish FOP association, Svenska FOP-föreningen that I founded in 2004. Today we accept members from all Nordic and Baltic countries. We are one of the members of Rare diseases Sweden since 2008. I am in the steering committee of the International FOP Associations (IFOPA) Presidents Council (IPC) where I have been a representative since the start of IPC in 2007. And I am involved in different multi stakeholder constellations for FOP on a European and Global level.

**Is something similar between OI and FOP?**
In a few words, the pain, and the not knowing what you wake up to. To handle with extra care in every situation. But even if you do that, you are not safe and secured from pain or more limitations. And there is also a complex balance between “safety and fun” in life.

**Anything that’s very different?**
FOP creates extra bone. These are perfectly healthy bones, but in the wrong place. People with OI have brittle bones.

**What do you think of the Stockholm conference?**
It was one of the best conferences I have attended (and that says a lot). I did not know what to expect from it before it started. It was so many short talks, jam packed, and first I felt “why so many talks”. But in the end, it all made perfect sense. To really get the holistic perspective of the rare bone diseases, to not focus so much on the bones, to focus more on what we can actually do to improve our lives with therapies and functional medicine. I learned so much about similarities between rare bone disease which I had no clue about before this conference.

**If you could suggest a topic OIFE and the FOP-community could work together on - what would it be?**
It is hard to pick one. I see so many topics, which is why I give you three:
1. To develop relevant questions in clinical trials to be able to measure the outcome of clinical trials from a rare bone perspective instead of a healthy perspective.

2. How can we handle the different kinds of pain we have in common with safe functional medicine to limit the intake of painkillers that come with bad side effects? And when do we need stronger medications and what to think about?

3. Nutrition and rare bone diseases. This has become my profession on the journey with FOP, to improve mental and physical health and make sure the body gets the best possible internal environment to better handle pain and medications and to limit the risk of unnecessary side effects from medications that can come from poor or unhealthy diets.

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**Flying OI Experts Nigeria**

OIFE’s support programme called ‘Flying OI Experts’ enables professionals to travel to less developed countries to teach and improve knowledge, treatment and social support in these countries. This can also lead to new initiatives in starting local OI-groups, that can provide support to more individuals and families.

We would like to express our gratitude to the two OI surgeons Dr. Aldofredo Santana (Venezuela) and Dr. Carlos Pargas (USA) for taking a week off their normally busy schedule and travel all the way from their home countries to Lagos, Nigeria as part of the OIFE Flying OI Expert programme.

The 2023 project was organized by Osteogenesis Imperfecta Foundation, Nigeria (OIFN) and financially supported by the OIFE and donations from members of the Norwegian OI-group NFOI. But without the dedicated doctors who volunteered to teach advanced OI surgery and do educational activities to increase the knowledge on OI in Nigeria – it would not happen. Thank you so much!

Read more about the Flying OI Experts programme [here](#). You can donate to the project [here](#).
Tell us a bit about yourself!
I'm 27 years old and I've been a stand-up comedian for eight years. Little by little I am better known in Spain (Television, radio, social networks...) and I have many followers on Instagram. Also, I just wrote a book, 'Being different is not so strange', where I talk about my life with OI, about disability, and I explain my personal vision of comedy.

How do you live?
Since a year I have been living with a roommate in a central apartment in Madrid. It has an elevator to the apartment, and I have also bought a small stool to reach the high furniture. I also have an adjustable bed. I normally use a cane to walk, although I also use a wheelchair for long distances.

In what way has OI affected your art?
In my comedy shows I talk about my height and my physique openly. Also, I'm talking about the OI. For example, I explain what OI is. I usually say "It means brittle bones. Meaning if I die, throw me in the recycling bin!" However, OI has never stopped me from doing what I like. I have done stand-up comedy, since I was 18 years old. I have even done shows sitting in a wheelchair due to a fracture or with casts.

What projects are you currently working on?
I am currently in full promotion of my book 'Being different is not so strange' and touring my country with my stand-up comedy show 'Hate comes in small bottles'. Soon I will release a new stand-up comedy special and a sketch comedy show. Maybe in the next few months I'll release a podcast, but who knows?

Why do you do what you do?
The first time I got on stage was to do poetry. Then, one day a friend told me that I was very funny and then I decided to try stand-up comedy. Now I do it because it's my lifestyle and it pays the bills. Also, doing stand-up is what makes me happiest.

What kind of work do you most enjoy doing?
I have always hated office work and work that forces me to get up early. Another advantage of dedicating myself to doing stand-up comedy.

What's your scariest experience related to your work?
There have been and will continue to be bad shows, but I especially remember the first one I did eight years ago. That was terrible! My first performance was for the benefit of deaf and mute people. I had the sun in my face, and I couldn't see the audience, and next to me there was a sign language interpreter who translated everything I said. Plus, it was in a school, on the street, with fairground rides playing. I thought the show was going very badly because I didn't hear anyone applauding. But it turns out that deaf and mute people do not clap their hands together, but rather they applaud by waving their hands in the air. But I didn't see them because I had the sun in my face. A real disaster.
What's your favourite artwork?
Stand-up comedy. I imagine it won't surprise anyone... Although I miss writing and reciting poetry.

What role does the artist have in society?
I believe that an artist has the duty to reflect to society what society itself does. Its defects and how to correct them. Its advantages and how to defend them. Furthermore, specifically in stand-up comedy, you have to be critical and try to make the limits of humour less rigid each time. In the specific case of artists with disabilities, I think it is an unbeatable opportunity to make people understand that, above all, we are people. We are not just a wheelchair, or a disease, or a cane...

What is your dream project?
I have been very lucky because in recent years I have been able to fulfil the dreams that I have set for myself. Lately I have a new one... Record a comedy special and have it distributed on a streaming platform like Netflix or HBO.

Do you have any messages for readers of OIFE Magazine or for OIFE?
The only message I can convey to you is the importance of humour, always. To heal, to be better people, to assert ourselves as people with disabilities, to have better mental health and to know ourselves. Let's never stop making jokes. And you know... See you at the recycling bin!

New resource for adults with OI

Check out the new resource from our friends in the USA, which in our opinion must be the best thing since sliced bread:

"The Adult Health Toolkit" made by the OI Foundation. It includes Information for Adults Living with OI, Their Families, and Medical Professionals to help you navigate the many aspects of managing your health as an adult living with OI.

Learn more and check out this resource at www.oif.org/adulthealth.
News in brief

OIFE EVENTS CALENDAR
Do you find it difficult to keep track of all the OI-related meetings and events? Did you know that OIFE keeps track of an events calendar, which includes OI-events, scientific events and other conferences relevant for people who are interested in OI? You can find it here.

GET TO KNOW EUCAPA!
EUCAPA aims at providing patients, and patient experts, with the adequate knowledge and skills to effectively participate in the Health Technology Assessment (HTA) process. Their courses are designed to empower participants to advocate for themselves and make their voices heard. This will be needed when future treatments for OI comes up for approval on a national level with HTA authorities or payers (cost benefit and budgets). Read more on the EUCAPA webpage.

BOOK ABOUT OI - NOW IN ENGLISH
Researchers from the Share4Rare project in Spain published a book about osteogenesis imperfecta (OI) some years ago. Now it's available online (for free) in English language as well. The Spanish organization Ahuce has provided input to the book. You can access it by clicking here.

EURORDIS PHOTO AWARD
The EURORDIS Black Pearl Photo Award 2024 is now open! Every year, hundreds of people from all around the world submit their photos to our photo award. The contest is an opportunity to visually express what it means to live with a rare condition and to share your story with the rare disease community and beyond! People with OI or their families have won or been in the top 3 several times. One of the winning photos featuring OI you can see below. Can we do it again? See more information here.
NEW PHD IN OI – DENTIST
We congratulate dentist Clara Garcete Delvalle DDS. Msc. from Spain with her recent PhD in dental treatment of osteogenesis imperfecta!

NEW PHD IN OI – EXTRASKELETAL ISSUES
We congratulate Dr. Silvia Storoni from the Netherlands, with her recent PhD! The thesis is called "Osteogenesis Imperfecta - new insights into extraskeletal complications": http://doi.org/10.5463/thesis.416

Several of the articles are focused on pulmonary issues in people with OI, which is a very important topic for the community. We look forward to hearing more from Silvia in the coming months and years.

EU DISABILITY CARD
In September the European Commission launched its legislative proposal for expanding the European Disability Card across the EU, to ensure access to a range of services under preferential conditions for EU citizens with disabilities. See more information here. EURORDIS welcomed the proposal as an "important step in the right direction", but not without also noting that the proposal "should have been more ambitious". Read the statement from EURORDIS here.

PERSONALITY & PAIN IN OI
Can your personality influence the way you experience and deal with pain? This is what OI researcher Ruben Muñoz Cortés, tried to find out by analyzing survey answers from a large number of adults with OI. Now the article has been published: https://tinyurl.com/h7u7zp9p The topic was also covered in Ruben's talk in Stockholm.

SEXUALITY AND OI
The social worker of Fundación Ahuce helped organize an event called "Sexuality, adolescents and rare conditions" together with the organization Sexualidad y Discapacidad (Sexuality and Disability).
Physician statements on children with severe types of OI

Dear colleague,

In the literature there is very limited information on fracture incidence during the first years of life in children with OI type III and IV on early bisphosphonate treatment. We understand your busy schedule, but we would appreciate your quick input on the statement below. Please only answer if you are a physician who meet children with severe types of OI. The information will be used in the preparations of analysis of fracture rate in the BOOSTB4 trial. The data will be handled anonymized. Please mark your opinion in the table (you can contact us if you want the form sent to you by email).

Thank you very much in advance,
The BOOSTB4 team
www.boostb4.eu
boostb4@clintec.ki.se

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<th>No</th>
<th>Comment, if any:</th>
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<td>3</td>
<td>Approximate age when you start rodding surgery?</td>
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<td>4</td>
<td>Years of experience caring for children with OI?</td>
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*with bisphosphonate treatment starting approximately before 6 month of age and with standard of care as of today.

Cecilia Götherström | Associate Professor
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https://staff.ki.se/people/cecgot
A multicenter phase 3 clinical trial is being conducted to study the efficacy and safety of a clinical trial drug compared to bisphosphonates in children and adolescents who have OI. The trial is open to eligible children from age 5 to less than 18 years of age who have a diagnosis of OI Type I through IV. The clinical trial will have sites participating in the following countries: Canada, United States, Australia, Japan, Austria, Belgium, Hungary, Poland, Slovakia, Switzerland, France, Germany, Spain, Turkey, and United Kingdom. For more information, please visit the clinical trial registry page at: https://clinicaltrials.gov/study/NCT05972551.

Ultragenyx-led ORBIT Study
Ultragenyx, in partnership with Mereo, are leading the Orbit clinical study, which is for individuals living with osteogenesis imperfecta (OI) Types I, III, and IV. The purpose of this study is to investigate the efficacy and safety of setrusumab, a monoclonal antibody, for the potential treatment of OI in pediatric and young adult patients. The study aims to understand the potential reduction in fractures as well as other impacts of OI. Study participants are at least 5 but not yet 26 years of age, have a confirmed diagnosis of OI Types I, III, or IV, and are willing not to receive bisphosphonate therapy during the study. Learn more about this study here or visit https://www.ultraclinicaltrials.com/OI.

Ultragenyx-led COSMIC Study
The purpose of this study is to evaluate the effect of setrusumab, a monoclonal antibody, against intravenous bisphosphonates (IV-BP) in children living with types I, III or IV OI. It is also led by Ultragenyx, in partnership with Mereo. The study is focusing on reduction in fracture rate, including morphometric vertebral fractures, in younger paediatric participants as well as other parameters.

Currently enrolling patients aged 2 but less than 7 years old with OI Types I, III, and IV. Learn more about this study here or visit https://www.ultraclinicaltrials.com/OI.

Energy4OI Fitness app
Energy4OI is a project in which Care4Brittle Bones wants to boost the physical well-being of people with OI. This is done through a self-practice app, designed by and for the OI community together with professionals who have supported OI for many years. From January 1, 2024, they will start a project for everyone with OI aged 18 and older who wants to work on their physical well-being.
For this project Care4BrittleBones invites more than 100 people to use the “ENERGY4OI app” with the aim of improving the quality of life in people with OI by getting them to exercise more in a safe and responsible way.

The pilot project is organized by the Care4BrittleBones Foundation together with Dr. Antonella LoMauro, Politecnico, Milan, Italy. The project leader is Sander Colijn, a young doctor with OI, based in the Netherlands. If you are interested in participating, please check out more information and register here and we will get back to you for a more formal intake: [https://www.surveymonkey.com/r/ENERGY4OI](https://www.surveymonkey.com/r/ENERGY4OI)

**“Puppet theatre” study – “The Hospital of No Surprises”**

The “Puppet theatre” study is a McGill ethically-approved research study aimed at optimizing the surgical experiences of children with Osteogenesis Imperfecta. The purpose of this study is to professionally produce an animated version of a children’s play written by one of our research assistants/artists titled “The Hospital of No Surprises”, and to create an accompanying educational toolkit in collaboration with the global OI community (children, parents, and key stakeholders).

**For parents and key stakeholders:**
Researchers at McGill University are seeking expertise from individuals who are affected by Osteogenesis Imperfecta (OI)!

- Do you have a child with OI or have OI yourself?
- Are you involved in the OI community or provide care to children with OI?
- Are you over 18 years of age?

If you answered YES to any of these questions, you may be eligible to participate in our study to understand the experiences of children with OI and improve care. Contact a member of the research team today!

**For children:**
Researchers at McGill University are looking for advice from children affected by Osteogenesis Imperfecta (OI)!

- Are you under 18 years of age and have OI?
- Do you have a sibling with OI?

If you answered YES to any of these questions, you may be eligible to participate in our study! We need your help to understand your experiences in the hospital and make the hospital better for others like you. Contact a member of the research team today!

**For more information, please contact**

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Email: Argerie.tsimicalis@mcgill.ca
Future Events

The Quality of Life 4 OI conference is organized by Care4BrittleBones and provides a platform to engage on clinical practice and research for OI. It will be examining ways to measure Quality of Life in all areas of OI research and care to improve outcomes for people with OI. The event welcomes clinicians, researchers, allied health professionals, industry representatives as well as the OI-community.

More info at www.qualityoflife4OI.org

OIFE Calendar
For an updated list of events & conferences - see OIFE's web calendar: http://bit.ly/36A6mw8

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Ute Wallentin & Maria Barbero (Coord. Social Network): socialnetwork@oife.org
Stefanie Wagner (newsletter editor and secretary): secretary@oife.org
Youth Coordinators: youth@oife.org

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X (Twitter): @OIFE_OI
Instagram: https://www.instagram.com/oioife/
LinkedIn: https://www.linkedin.com/company/oife