Editorial

By Ingunn Westerheim, OIFE President

WHAT DOES OIFE MEAN TO YOU?

Yesterday we received the first version of the after video from “Balancing Life with OI”. We cannot wait to share it with the rest of you. One of the questions that was asked in the video was “What does OIFE mean to you?”. This can be a tricky answer to answer sometimes, because OIFE is not an ordinary national patient organization. OIFE is an umbrella – with a complex set of activities and goals. And it can be difficult to describe who we are and what we do. Especially for our national delegates, when their local members ask.

The easy answer for me personally, would be to say that OIFE is my baby. I’ve been the OIFE president for eight years now and I have seen the organization grow and develop with the help of countless engaged and dedicated volunteers. OIFE is very close to my heart and every day I think about how we can develop and strengthen the umbrella organization even further. Not alone, but with the help of our many volunteers – existing and future.
OIFE is also friendship. Both in OIFE AGMs, topical meetings and OIFE Youth Events – friendships for life have been established, just like they do in national events.

A national OI-organization is truly a unique space, where people with the same appearance, experiences and mindset can come together to laugh, share, create and work to develop better services and treatment for OI in their countries. Or just to enjoy the presence of peers.

Or we can meet on a European level as we do. In co-existence with our good friends and contacts outside Europe, who provide us with good ideas, best practices, and perspectives. And the latter is perhaps the most important. Jesus - the leader of the newly re-established OI-organization in Panama, recently told me that a baby girl with OI in Panama passed away because of pulmonary issues. Maybe she would have survived in another country with better care and more resources? Maybe not. But it’s a fact that a lot of people in different corners of the world put in a lot of energy, to avoid that babies and adults with OI die – because treatment and knowledge of OI is not good enough in their country.

OIFE is also network. We are a huge international community with strong collaborations between researchers, clinicians, industry representatives and people with OI and their families. On a relatively equal level.

In Stockholm we all came together face to face, to exchange ideas, knowledge, and experiences on living with pain and OI – and how we can improve assessment, measurement and not to forget, management of pain for children, adolescents and adults with OI. For me the conference was a memorable experience. We had worked for almost 2 years to make it happen. And to see and hear so many interesting and informative talks, anonymous testimonies and people sharing from their own experiences and knowledge was amazing. We are working hard to edit and text the recordings from the conference – to make it available to all of those who did not have the opportunity to be there – either because of time, travel, money or health reasons.

OIFE is also knowledge and trust. More and more often stakeholders contact us to hear the voice and perspectives of people with OI. This includes researchers who want input for their research applications, companies who are planning their clinical trials or clinicians who want to know how they can offer better services to patients with OI. Sometimes I feel we are getting too popular. Because we struggle to meet the demands. But this is a luxury problem. It only means we need to recruit more board members, train more patient experts and motivate more and younger people to become involved as volunteers for OIFE and the national OI-organizations.

Because sometimes we tend to forget – without the national OI-organizations, OIFE is nothing. We are not a national organization with individual members. We are an umbrella consisting of 40 member organizations, and without them we cannot do a lot. We have 20 ordinary members in Europe, 15 national organizations outside Europe and 5 supporting members, where CLAN was the latest addition. You can read more about them in this magazine.

I wish you a very good summer and autumn. See you in Valencia next year! Maybe we will meet some future volunteers there?

Kind greetings from
Ingunn – OIFE president
What is the OIFE doing?
By Ingunn Westerheim, OIFE President

Between May and September, life in OIFE has been before and after the Stockholm conference. A lot of energy went into the preparations of the topical meeting ‘Balancing Life with OI’. The conference was followed by our annual general assembly, which perhaps wasn’t as well prepared as it could have been. But the conference was a huge success – with more than 200 participants if you count both those who were face to face and those who followed on streaming. A remarkable 98% of those who answered the evaluation, said that the conference met their expectations. So, either people had low expectations, or we did something right. After some less busy weeks, we have been picking up the activities in the IMPACT project and the Pain Project again. We have also been busy planning events like the Youth Event, the OIFE Clinical Trial Update and the 2nd OIFE Investigator Meeting.

In addition to the big event, we had many smaller meetings with clinicians, researchers, companies and organizations, especially connected to clinical trials, the Pain Projects and future events. These are some of the meetings we have organized or attended between May and September:

- OIFE Board Meetings (Zoom), May 16, May 24, June 5, June 20, July 11, Aug 8
- XLH Alliance, May 8 and July 10
- OIFE Drop In session May 30
- RBD: Mobility SLR and Manuscript, Kick-Off call, June 2 (IW)
- AI and patient voice (Semantic Hub), June 29 (IW)
- An EU Ecosystem for Rare Diseases Conference, June 29 (CF)
- IMPACT Steering Committee, June 16 (IW and TvW)
- EuRR-Bone Study Group Meeting, June 3 (IW, TvW, RTS and CF)
- ECTS Webinar Case Studies OI, June 5 (IW)
- Sanofi, July 18 (IW)
- Relation Therapeutics, Aug 16 (IW)

THE PAIN & OI PROJECT
We’ve organized two workshops (resource group) in the Pain and OI Project this year. But the main happening in 2023 was of course the topical meeting “Balancing Life with OI” in Stockholm. Since we didn’t want things to end with the conference, we are working in different ways to disseminate the knowledge and results. We have made an after movie and we are editing and texting the recordings of all the talks we have permission to share. We will publish them on YouTube as soon as they are ready.

We are also writing up a more formal report from the conference in addition to some recommendations, which we will ask the expert resource group to endorse. We have also hired a person who will help us develop a toolbox. In addition, we are contributing to a scientific article with the main results from the pain and OI survey, which will be written by experts from Baylor College of Medicine. We have also provided input to other pain related projects, like the pain module in the Teen and OI project of Shriners Hospital in Montreal and we’ve also provided advice from patient perspective in the Pain project of Sanofi. See https://oife.org/what-we-do/pain-oi/

AFRICAN OI EVENT
On May 27 the OIFE was represented at the international African networking event hosted by the Osteogenesis Imperfecta Foundation Nigeria. The purpose of the event was to provide information, but also inspire more collaboration between formal and informal groups in Africa.

![Balancing Life with OI/disability International Awareness 2023](image)
EURORDIS EVENTS
On May 17th our Board member Malene represented OIFE at the general assembly of EURORDIS, which took place online. And also this year, people connected to OIFE were attending EURORDIS Summer School in Barcelona. Physiotherapist Miguel R. Molina took part on behalf of the Spanish organization Ahuce. Since Miguel is also a member of OIFE’s Medical Advisory Board - OIFE will also benefit. Check out this webpage, if you are interested in learning more about the educational opportunities that the Open Academy from EURORDIS can offer: https://openacademy.eurordis.org/

The 26th and 27th of May 2023, OIFE attended the EURORDIS Membership Meeting in Stockholm. More than 200 participants attended the conference. The workshops were split into different phases of life; Children, Adolescence, Adults and Aging. All of which had an overall emphasis on a holistic approach. OIFE delegate Rebecca Skarberg from Norway was the moderator for the sessions on adults, and Lars Nesset Romundstad, deputy chair of NFOI represented OIFE as a speaker in a panel discussion about the different aspects of adolescence and rare diseases. From the OI community, Inger-Margrethe Paulsen also attended, representing the Norwegian OI association. Read a full report from Lars here: https://oife.org/2023/06/16/oife-at-eurordis-meeting/

A UNION OF EQUALITY
On June 27 OIFE volunteer Rebecca T. Skarberg represented EURORDIS at the EU Meeting “A Union of Equality: Disability Rights and Strategies”, which was a seminar on examples of how to reach equality by using strategies to realize the rights of persons with disabilities. In the EU Disability Platform, Rebecca is a member of a subgroup on Independent Living. Their mandate is to find out how many disabled people who are living in an institution or a group home. Different interpretations on how the EU countries define “independent living” are being discussed. Read more: https://oife.org/2023/08/03/a-union-of-equality/
34 percent of all individuals with OI report that pain has influenced their career choices, and 30 percent also state that pain affects their social life. - This is just one of the many facts we learned during the OIFE’s Balancing Life With OI conference in Stockholm at the beginning of June.

I had arrived in Stockholm ahead of time and had a day and a half to explore the hilly streets of Stockholm before the conference began. As a result, at 2:30 the night before the conference, I found myself lying in bed with sore shoulders, unable to sleep. Realizing I needed some rest, I had to leave the hotel and roll across the street to a 24-hour pharmacy to buy a pack of paracetamol. There is perhaps no better way to prepare for a conference whose main theme is pain!

The conference spanned two days, featuring a packed schedule with speakers taking the stage one after another with 10 to 15 minutes in between. Facts and information were delivered like jabs in a boxing match, leaving attendees both wiser and somewhat dazed.

**Definition of pain**

The first speaker on the program was Audun Stubhaug, who delved into the nature of pain. He used the following definition, also employed by the International Association for the Study of Pain - IASP:

"An unpleasant sensory and emotional experience associated with, or resembling that associated with, actual or potential tissue damage."

The truth is, we all know what pain is when we feel it, but there can be significant individual differences in how we experience it. He referred, among other things, to a Danish study that examined how patients experienced the exact same type of operation. Some felt almost no pain, while others reported experiencing maximum pain.
At the same time, he emphasized the differences in how acute pain and chronic pain should be treated. The goal with the former is to eliminate the pain during the period it is felt. Acute pain is often measured in hours or days and can easily be treated with anything from paracetamol to powerful and fast-acting opioids. In contrast, the treatment of chronic pain should aim not only for pain relief but also for improved function and long-term quality of life for the patient.

**Causes are complex**
Mercedes Rodriguez Celin, a pediatrician at Shriners Hospital in Chicago, also referred to a study involving 861 participants with OI, where 42 percent reported experiencing chronic pain. The causes of pain among people with OI are often complex, she explained. It can be orthopaedic, such as fractures and deformities, but it can also be related to muscle weakness, unstable joints, poor alignment, or neurologic and inflammatory issues.

Gastrointestinal problems are commonly observed among individuals with OI, and there can be various causes for them. Factors such as body proportions, with many having a small upper body, as well as scoliosis and chest deformity, which leave less room for the stomach and intestines, can lead to symptoms like stomach pain, acid reflux, and constipation.

However, tissue-related issues or factors related to diet and activity level can also contribute to the gastrointestinal related pain. Increasing focus on diet and activity can therefore help alleviate these problems, as explained by Lena Lande Wekre.

**Pain is the most common symptom in OI**
OIFE’s own Impact Survey also reveals that among the symptoms reported by individuals in recent months, pain is the top scorer. Out of the 1,442 participants, 1,181 experienced pain in the past year, while fatigue, soft tissues, and scoliosis followed suit.
Many also reported experiencing sleep problems, but Marie Coussens from Ghent University in Belgium conducted a study where 35 OI patients were equipped with an accelerometer to measure their sleep quality. The study overall showed a reasonable amount of sleep, with participants averaging 7 hours and 49 minutes. However, the sleep study also revealed that OI patients were awake for an average of 45 minutes during the night, significantly more than other individuals.

**Touching testimonies**
Many of us are experts in living our lives despite limitations, and this was evident at the conference through a series of touching anonymous testimonies that were read out.

I had the pleasure of speaking with American Karen Braitmayer about balancing work, ambitions, and pain, while Kis Holm Laursen shared her experiences of parenting a child with pain, and AnnBett Kirkebæk talked about the impact of pain on relationships and sex life.

Even though we have learned to cope, many still report that pain has a negative impact on their physical lives, leisure activities, and daily tasks. On the other hand, studies show that individuals with OI only experience a minor impact of pain on their ability to lead independent lives and on their close relationships with family and friends.

*If you were unable to attend the topical meeting "Balancing Life with OI" in Stockholm, you can download the abstract book with summaries from all the speakers for free at our webpage: [https://tinyurl.com/3mye6jhz](https://tinyurl.com/3mye6jhz)*
Impressions from Stockholm

Alessandra Tolaccia

My name is Alessandra and I am 48 years old. I have OI type 1. In my early 30s I started experiencing pain which over time became chronic and it started effecting many aspects of my life.

From my point of view the conference was perfectly organized and everything run smoothly with many extremely interesting presentations and a rich program. There was also time to meet with fellow patients and the lecturers which was a plus.

My personal highlight: The fact that for the first time, to my knowledge, there has been so much focus on the pain issue on adults with OI. It has been extremely interesting to see how all the studies that have been presented showed that people with OI type 1 are the ones within the OI community that are experiencing more chronic pain. This “new” interest in pain and pain related issues is something that gives me hope for future.

Dr. Cecilia Jimenez-Moreno

I attended the virtual version in representation of the team, and I really enjoyed it. It was very much science driven and the quality of the talks was high. You made me want to go back to my academic life! I took lots of notes, both from the talks but also from questions raised by the audience (as these are for me as informative as the response) and I will be sharing them with the team on our next call.

You also did an amazing job intercalating patients’ voices and testimonies; then, the emotional wave was not all at once and you kept reminding the audience the why of all that research. So, in summary, hands up to you and OIFE and thank you so much for chasing up my registration so I couldn’t miss it.

Eva Sacherer

My name is Eva Sacherer from Austria. I am the OIFE delegate for the Austrian OI organization, called OIA. I am mother to my beloved son Jakob who just turned two years old in May.

The amount of information in Stockholm was overwhelming to me. On the one hand it was kind of practical to get very short snaps of inside views on different topics. If this or that topic was the one you were interested in, you could talk to the presenters afterwards during coffee breaks. You could tell that there was a lot going on in this field around the world.

I had been waiting all weekend for an explanation as to why Jacob gets spasms when his femur breaks. He suffers a lot from it, and I was looking for a way to take this acute pain away from him as quickly as possible. I was surprised that nobody mentioned it.

As luck would have it, a paediatric surgeon sat down next to me during the gala dinner. After some small talk about the fishy food here in Stockholm, the sound of music (foreigners always mention this to Austrian citizens) and the medical care given to the children in the USA, I asked about the spasms and received both this and Jakob’s overall Habitus answers. Arnica, Arnica, Arnica tinctures and immobilize the leg.
as much as possible, sometimes cool, sometimes warm, everyone is different, but provide effective pain medication as quickly as possible.

Stockholm impressed and moved me deeply. Pain and fear accompany both children and parents throughout their lives. If we still manage to make Jakob have a little less pain the next time he breaks, then it was worth the trip. Thanks, OIFE for this brilliant conference!

Dr. Frederik Heinrich
I am Frederik Heinrich, 43 years old, living in Berlin, Germany. I am a scientist working as a bioinformatician at the German Arthritis Research Center Berlin (DRFZ), that focuses on understanding the immune system in health and disease. One of the main emphasis lies on autoimmunity such as rheumatic diseases. I have OI myself, and my parents were cofounders of the German OI foundation almost 40 years ago.

I really enjoyed the Stockholm conference. I highly rejoice international gatherings, because it simply widens one’s horizon. Hanging out with other OI people to share common experiences is always a bit of a vacation from everyday life, but doing that in an international context, is even greater.

Talks, and people presenting at the conference were very interesting, the conference schedule was pretty tight, but very well organized. Maybe a bit more breaks in between, or less presentations all together would be good. At a certain point I always kind of get saturated with information I can conceive. The place was really great, and I liked the bit of Stockholm, which I explored, a lot. My highlight was definitely meeting OI people from around the globe, and getting together, and simply having a good time.

Marie Coussens
I am a physio-therapist and in the final stages of my PhD at Ghent University (Belgium). I have a particular interest in adults with hereditary connective tissue disorders such as OI. My research focuses on the identification of bone and muscle issues (e.g., muscle weakness, altered body composition, lower resistance of bones to external loads, the reactivity of the bone to muscle strains) in these individuals.

Based on these findings, we aim to set up a safe training which may improve both some of these bone and muscle issues in individuals with OI.

It was the first time I joined an OIFE conference, and it was really enriching for me. The Stockholm conference gave the opportunity to gain insight in the perspectives of not only researchers involved in OI research, but also from the OI community and clinicians working with individuals with OI.

Also, it highlighted the importance of pain in individuals with OI and the high need for further research and appropriate treatments. As a researcher, these perspectives provided me with further insight in the current research priorities and may as such further guide the direction of our research.

My personal highlight: The solidarity among people with or working with OI.
OIFE Annual General Meeting in Stockholm

On June 10th and 11th 2023, we came together for OIFE’s Annual General Meeting (AGM). This time we met, in real life, in Stockholm, Sweden. There were approximately 40 people representing 15 ordinary (European) and 4 associate member organizations (incl. USA, Peru, Ahuce Foundation and Care4BrittleBones).

On Saturday afternoon we took care of the formal business and the agenda included:
- Internal news & updates
- Approval of OIFE’s Annual Report 2022
- Approval of financial report 2022
- Elections and appointment of volunteers
- OIFE long term strategy

Ingunn went through the highlights of the annual report and explained about activities held in 2022. Coreen explained about the finances including why we have 2 different financial reports for 2022 (moving to Belgium) – and that they need to be evaluated together. Both the annual report, the financial report (part 2 of 2022) and the budget were approved unanimously by the voting members.
Unfortunately, due to time constraints, the Board had not managed to finish the entire draft of the new long term strategy. But we agreed on a set of new goals and the members gave the Board the authority to continue the development of the strategy according to these overarching goals. Elections were held and OIFE welcomed a new Board member Jacob Ø. Wittorff from Denmark.

On Saturday evening, we had a nice conference dinner together with the members of the Swedish organization SFOI, who had a separate meeting in parallel with the OIFE AGM. The dinner also included some invited speakers who needed to stay until Sunday. During the dinner we did a toast to the OIFE’s 30th birthday, which took place in March 2023.

On the Sunday we originally planned to do workshops on various topics. But we changed the plans and did a plenary discussion about OIFE priorities and future events. We also did an evaluation of the topical meeting. The feedback from the OIFE members was good, but many thought the programme was too packed and were missing breaks to digest the information and socialize with other participants. We agreed to do next year’s AGM online, to make it possible for associate members to attend. From September 5-8th 2024, we will do our annual conference for our member organizations in Valencia, Spain, which will focus on organizational development, advocacy and patient involvement.

Meet the new OIFE Board Member

JACOB Ø. WITTORFF, DENMARK
My name is Jacob, and I’m a new member of the OIFE-board and also member of the board of the Danish OI Organization, DFOI. I have OI type 3 and was born in a small town in southern Jutland, Denmark, 42 years ago. However, I have been living in Copenhagen for the last 18 years.

I consider myself relatively new to the OI community. As a small child, I attended a few OI meetings with my parents, but I haven’t really been part of the community until shortly before the pandemic. My relationship to OIFE began during lockdown when I participated in the OIFE Virus Workout sessions on Zoom.

During my teenage and young adult years, I was focused on appearing "normal," hanging out with friends without disabilities, and I didn’t want to dwell too much on my disability. Looking back, I realize that I may have missed out on a lot by not being part of the community and meeting peers my own age who face similar issues.

What do you do when you’re not doing OI-work?
I work as a journalist for a Danish tech publication, where I cover the tech industry and how technology impacts business and society. When I’m not working or involved in OI-related activities, I enjoy attending football matches. I’m a proud season ticket holder of the Danish football team, Brøndby IF.
I also like going to museums, eating out, and one of my favourite activities is going on long rolls in my wheelchair. It's great exercise, a fantastic stress reliever, and those trips make me feel most independent. Unfortunately, there are times when I roll longer and more frequently than my body appreciates. As I get older, I'm more cautious not to overdo it and try to mix it up with other types of exercise.

In your opinion, what is one of the most important jobs/tasks/areas for OIFE?
Knowledge sharing about medical treatment and research is, of course, crucial. But I also believe that OIFE can facilitate many important discussions about some of the more uncomfortable issues related to living with OI.

Meet the OIFE Delegates – Associate Members

TED TRAHAN, USA
My name is Ted Trahan. I am married to Sharon, and we have two adult children, Greg and Käthe. Greg has OI and we are excited to share that he is recently engaged to Natalie, who also has OI.

In what way does OI affect you personally?
As a parent of a child with OI, osteogenesis imperfecta has presented me with the highest highs and lowest lows. The lows come from watching my child endure the effects of OI and associated surgeries, while the highs come from watching him and others in the OI community overcome barriers to lead their lives and just become great people!

The OI Foundation
I am on the board of the U.S. Osteogenesis Imperfecta Foundation (OIF) and am currently serving as President. The OIF started with a story Gemma Geisman wrote to Redbook Magazine in 1967 about her experiences raising a son with OI. Following publication of that story, letters poured in from families in similar situations from all across the nation. Due to that response, Gemma and several other parents founded the OI Foundation in 1970.

The most important work of the OIF is stated in our mission: to improve the quality of life for those living with osteogenesis imperfecta through research, education, awareness, and mutual support.

Today the OIF reaches more than 10,000 individuals and families of people with OI through social media, in addition to hundreds of individuals signed up within its membership. OIF programming is coordinated by staff, with local and regional events run or supported by volunteers. In addition, the staff is responsible for oversight of information and resources for families and medical professionals, scientific and research meetings, national conferences, and liaisons with other patient advocacy groups.

We are eagerly looking forward to the OIF’s first in-person national conference since the COVID-19 pandemic. The conference will be held 19-21 July 2024 in Omaha, Nebraska.

What do you do in your free time?
Outside of the OIF and work I like to sail. Sailing is a crew activity. As captain I rely on a competent first mate and a crew that takes care of tasks and provides camaraderie. The same is true for us individually, with or without OI. Develop your first mate and crew to support you as you captain your own life.
ISIS ATUNCAR, PERU

My name is Isis Atuncar, I am from Peru, and I was diagnosed with OI when I was born in 1986. I had the opportunity to receive medication and surgical treatment in the USA. That treatment improved my quality of life. When we were back in Peru, my parents and other families together formed the OI Association from Peru, with the aim of raising awareness about the condition and providing support to more affected families.

How does OI affect you personally?
The condition hasn't affected me much due to the treatments I've received throughout my life. However, in Peru, barriers exist in terms of mobility and accessibility, making it challenging to lead an independent life. Nevertheless, with the support of my parents, I've been able to pursue education and work in the social sector.

Tell us about OI Peru!
The Peruvian OI Association was founded in 1994. We have 200 active members, but we estimate that there are around 2000 patients in Peru. The association is led by a board of directors consisting of OI patients and their family members. Among the projects and activities, we carry out, there are:
• Providing support in managing medical treatment for patients with limited financial resources.
• Advocating with health authorities to ensure treatment is provided in hospitals across the country.

It is essential to ensure comprehensive treatment for OI patients throughout the country. It's important for OI patients to have a place in society, and with the support and tools we provide, they can achieve personal, professional, and educational development.

What do you do apart from OI work?
Apart from my work with OI, I study a master's degree in Spain, and I am an English teacher, I teach kids and adults. In my free time, I practice swimming, read books, and watch films.

We welcome the new OIFE Member CLAN!

Interview with Téa La, Community Development Officer for CLAN (Caring & Living As Neighbours)

At OIFE’s Annual General Assembly on June 10th, we were happy to announce that the OIFE Board has accepted the membership application from the NGO CLAN (Caring & Living as Neighbours), to become a supporting member of OIFE. CLAN is an Australian-based, not-for-profit, non-governmental organisation (NGO).

Tell us a bit about yourself.
Hello! My name is Téa, I am the Community Development Officer for CLAN (Caring & Living As Neighbours). My journey as an intern for CLAN first began with a focus on the Osteogenesis Imperfecta (OI) community in Fiji, Korea and Indonesia, but my work has since expanded to include other chronic health conditions of childhood (including Congenital Adrenal Hyperplasia (CAH), Type 1 Diabetes (T1D) and other non-communicable diseases (NCDs)). The heart-warming passion of families and children, the dedication of health professionals, and support from CLAN and partners has reaffirmed my career aspirations in the physiotherapy industry.
Can you tell us about CLAN?

CLAN, a non-governmental organisation (NGO) headquartered in Australia, is dedicated to optimising quality of life for children living with NCDs (non-communicable diseases) and other chronic health conditions in resource-poor settings, so they may grow to enjoy healthy and happy lives. We work in a range of countries around the world and with many different cultures. CLAN is privileged to have collaborated with OI communities in Fiji, Indonesia, Pakistan, and the Philippines.

In Indonesia I have been actively involved in supporting FOSTEO, the Indonesian OI community. CLAN’s efforts are informed by our Strategic Framework for Action which centres children and communities as a visual hub of focus for collaborative action around our Five Pillars: affordable access to essential medication and medical equipment; education, research and advocacy; optimisation of medical management; encouragement of family support groups; and reducing poverty and promoting financial independence.

The support and work we do is informed by the local needs and priorities of the children, families, and communities we work with. Over the years, CLAN has been fortunate to receive support from our partners and sponsors, and their donations continue to help us achieve our mission to redress health inequities. More information about CLAN and our Strategic Framework for Action can be found on our website.

How does CLAN benefit from OIFE as a member?

We are honoured to join OIFE as a supporting member. International collaboration, such as this, is crucial to effecting change for children living with OI. By bringing our experiences and resources together, we are better placed to strengthen communities and drive change to redress inequities. One example of this was our collective rights-based advocacy effort this year. With the amazing support from Jo Ragen and the International OI community, including OIFE, we were able to launch our OI Child-Friendly Rights Flyer on Wishbone Day 2023. The flyer supports OI advocacy efforts and helps children and young people living with OI better understand their basic human rights as declared under the United Nations Convention on the Rights of the Child. We are grateful for the collaborative efforts and support in the sharing of the flyer by OIFE. The flyer can be accessed here.
Can you tell us a bit about the situation of the children with OI in Indonesia?
The unprecedented COVID-19 pandemic and financial burdens placed on families had disrupted FOSTEO’s annual Wishbone Day (WBD) celebrations for 3 years. The FOSTEO community has expressed their desire to hold larger and more frequent events to both help strengthen the OI community’s bond and garner attention from the wider community to raise awareness. Affordability and accessibility of mobility aids and specialized health care remain key challenges facing families.

CLAN collaborated with FOSTEO in preparation of WBD 2023 celebrations in Indonesia. The organized event included an education seminar, Q&A session with specialist doctors, a fun painting session for the children, sharing session, and WBD-themed food and shirts. CLAN was proud to support FOSTEO in the event planning process, sharing of resources and the provision of a small grant to support their celebrations.

What are your plans for the future?
CLAN is proud to be part of the @MATES4Kids (Maximizing Access to Essential Supplies for Children) movement to reduce the preventable mortality associated with CAH by 30% by 2023, and contribute to efforts to achieve the United Nations’ Sustainable Development Goals (SDGs). By working with like-minded partners to develop a Community of Practice (CoP), champions from around the globe meet regularly to exchange knowledge, resources, and share experiences to scale action. More partnerships and funding to support these activities would help to further expand our efforts to reduce preventable mortality across other childhood NCDs such as OI.

CLAN continues to establish ongoing collaborative partnerships in efforts to optimize the quality of life of children living with OI, and promote awareness of their rights and needs. The OI Child-Friendly Rights Flyer (currently available in English, Indonesian and Korean) supports these advocacy efforts, and we look forward to working with partners to translate this and other resources into more languages in the future!

Watch Development Officer Téa La give an introduction to who CLAN is and what they do. The video was first shown on OIFE’s AGM on June 10th 2023: https://youtu.be/MzKh6EWne-E
Other rare bone conditions than OI

Interview with Liana la Forgia, volunteer and a member of the scientific board of A.C.A.R. Aps, the Italian patient association for Multiple Osteochondromas (MO)

OIFE has for many years had a project called "A stronger BOND between us". The goal behind the project is to further develop an informal network between different stakeholders working on OI and other rare bone conditions (RBDs). The project has several subprojects and activities. We are formal members of the European Rare Bone Forum (ERBF) and we have an informal long standing collaboration with the umbrella for XLH – the International XLH Alliance. Because some of the other rare bone conditions are very rare or very little known, OIFE can play a role in creating awareness and spreading information about these conditions, which very often have a lot of commonalities with OI. This is why we are starting a column about other rare bone conditions, where we will present a new condition, volunteer or organization each time.

Who are you and what is your relation to a rare bone condition?

My name is Liana la Forgia, I am Italian and I live in England. I am a volunteer and a member of the scientific board of A.C.A.R. Aps, the Italian patient association for Multiple Osteochondromas (MO), Ollier disease and Maffucci syndrome. I am also an ERN BOND ePAG Advocate and a caregiver.

In a few words - tell us about MO and how it's affecting lives?

Multiple Osteochondromas is a genetic bone disorder characterised by benign cartilage capped tumors growing on long tubular bones, flat bones and ribs. These tumors are called exostoses or osteochondromas. MO may result in limb discrepancy, short stature, abnormal development of bones, restricted joint movements, compressed nerves and blood vessels, pain and fatigue. MO is typically passed down from parent to child and it could also be the result of a spontaneous gene mutation. MO is characterised by the growth of osteochondromas which poses a risk of malignant transformation.

How rare is it?

MO is estimated to occur 1 in 50,000 people and is caused by a gene mutation. Surgery is currently the only form of treatment however, pain management may also help.

Which organizations are you involved in?

A.C.A.R. Aps stands for Associazione Conto Alla Rovescia, an acronym that acts as a countdown to finding a cure. A.C.A.R. Aps is the Italian patient association which supports patients and their families by providing guidance and information including listening to their needs, promotes and stimulates medical research to facilitate diagnosis and develop treatment aiming at improving the overall quality of care of patients. They also promote a collaboration of stakeholders and a multidisciplinary exchange between clinicians at an international level and raise the overall awareness also at political level of MO, Ollier Disease and Maffucci Syndrome.
Do you see any similarities or differences with OI?
To some extent, MO has several similarities with OI. For example, they are both genetic bone diseases, they both could also be characterised by limb discrepancy, short stature, mobility issues and chronic pain. Both patients with OI and patients with MO may have mixed emotions regarding their condition along with feelings of isolation. The sense of being considered “different”, as well as being burdened by fatigue, the potential impact of the condition on their social life, their work life, relationships, education and daily life overall, may all lead to poor self-esteem.

Anything that’s very different?
OI patients may often present bone fragility, a feature which differs from MO. Abnormal dentinogenesis, breathing problems and blue sclerae are not part of MO symptoms either.

What do you think of the Stockholm conference?
The Stockholm conference represented a great opportunity and an amazing experience overall. I am honoured to have been part of such a well-organised and significant event for OI and other bone disease communities. Many speakers presented their upcoming research which is often relevant to many other diseases, including rare bone diseases. Presentations were both inspirational and stimulating whilst being informative. The conference allowed me to learn a lot, I met many wonderful and interesting people, I had a chance to network but most importantly I boarded my plane back with a deeper understanding and a strong motivation to do more for the MO community as well as other disease communities I may come across in my role as a volunteer. Although the topic of the conference was “pain”, many other topics were touched and covered, all well-presented and engaging. The experience enriched me as a whole.

If there is one topic OIFE and the MO-community should work together on - what would it be?
The experience I had in Stockholm taught me that patients’ symptoms and experiences are not confined to one specific condition and that patients, caregivers and families have more in common with other disease communities than they are aware of. Joining forces, collaborating on common purposes and supporting each other is definitely the way forward. Together we may find answers and assist each other to face collective goals and issues, as well as identify ways to significantly help patients. I feel that the topic of “pain and fatigue” needs to be investigated further when considering these two conditions. I personally also feel that a better “quality of life” of patients and the “emotional impact of the condition on the inner self” may represent an opportunity to work alongside each other and collaborate together on common grounds. In particular, the experience of patient associations may prove useful for a “collaboration on designing natural history studies” for potential development of new therapies.
Meet Jay “Mini Producer” Manuel, a music producer and content creator based in Georgia, USA. Despite being hard of hearing and living with Osteogenesis Imperfecta Type 3 and Autism, Jay has dedicated his career to breaking barriers and representing young professionals with disabilities in the music and industry.

Please tell us a bit about yourself!
I am 27 years old, I’m currently living in Georgia, and I am an Entrepreneurship major. I am a full-time music producer and content creator. I’ve worked with thousands of independent recording artists from all over the world. I’ve created content for brands such as Coca Cola, Amazon, Snapchat, and more.

How does OI affect your daily life?
My OI and dependence on a power wheelchair impact my daily life, particularly when it comes to transportation. Networking online is more convenient for me, and going to events or business trips requires more planning. Additionally, I experience chronic pain, so I rest on worse days. I bulk record and schedule social media content to maintain consistency.

Can you describe a little where and how you live?
I live with my parents, grandmother, and fiancée, but we plan to move out on our own in the next year or so. Before the pandemic, I lived in an apartment, but moved back to my parents’ home when things got serious. I don’t drive because I don’t enjoy it and it overwhelms me. My parents drive me around and I pay them for it. I used to use transportation services, but they’re not available in our current area.

Are you active in an OI-organization - which and how? If no - why not?
I don’t participate in an OI organization because I’m busy with my music and content creation career, but I make content about my experiences with disabilities. The OIF has featured me on their Instagram before, and I’m now collaborating with companies and other creators to talk about disability-related topics.

What was the scariest situation in your life?
The scariest situation I’ve had is when I fell out of my chair while I was working on campus. I did have to ride in an ambulance and had to get surgery down the road for the arm that I had broken. I had also broken my femur as well.
What was the happiest moment in your life?
The happiest moment in my life is when I got to meet my now fiancée Pamela in person. It showed me that it was not only possible to travel by myself, but also that I could have a loving relationship with someone outside of my family or friends.

How did you discover that you wanted to become an actor and music producer?
I discovered that I wanted to be a music producer because I had a family member that had music equipment. This got me interested and I ended up getting my own software to start producing music in when I was in high school. Ever since then I've been creating beats and working with artists. When it came to becoming an actor, I always have seen myself being on TV and movies. Creating content has allowed me to explore what that looks like. I've done skits and even acted in various projects. I love what I get to do because every day is different.

In what way does OI affect your creative works?
The way OI directly affects my creative works is when it comes to my chronic pain. Some days are worse than others, so I have to rest which means I'm typically not working on anything. Also, OI affects how people perceive me in the world especially since I’m a little person as well.

Are there issues you must struggle with in your job due to your OI and autism? How do you compensate?
Even when I face obstacles due to my OI and autism. I’ve just learned how to adapt and overcome any given situation. That’s been the biggest key to my success so far.

Do you have a dream project?
I don’t have a specific dream project. I’m more so just excited to be experiencing the great things that life has to offer. I’m looking forward to being able to connect with more brands, individuals and be involved in opportunities that align with my morals and values. I love being able to impact others in a positive way and as long as I get to do that, I’m happy.

“Disability Burn-Out”: Internalized Ableism and its consequences!
By Raul Krauthausen, Disability activist

A disability can be stressful and sometimes trigger burnout. Often, one’s own internalized ableism plays a major role - in other words, the pressure of having to gain a raison d’être through performance and functionality. This can rule a lifetime.

It all started with a car accident. A crash out of nowhere, the great panic at the moment and then the realization that I had survived. But the "cure" is a tricky business, because it doesn't happen by itself; it is hard work that at times makes me despair.
For several months I was forced to deal almost exclusively with my body and with myself. I'm regularly in hospitals, and the longer I'm in there, the more diagnoses I get: Quadruple leg fracture, sleep apnea, hearing loss.

**I FEEL LIKE I'M IN A RABBIT HOLE**

A branching tunnel that seems to have no end. I notice how I digress from the starting point of the broken leg and deal with more and more new diagnoses. What threatens me at the end of this cave passage? Disability burnout, a state of physical or emotional exhaustion caused and exacerbated partly by my disability and partly by my internalized ableism.

That’s not an unrealistic expectation. The warning signs are already there: I'm getting tired faster and faster. I notice this in my increasing difficulty concentrating or setting priorities. I can't get going anymore and I'm moving in a downward spiral of disability, illness, exhaustion, inactivity and my own reproaches. So my condition is getting worse and worse.

**My body is not so robust and resilient as a body without a disability**

Two points play a particularly important role. My body and my mind. My body has a disability and because of this disability, the accident I experienced was life-threatening. I am light in weight and have fragile bones. The only reason I survived is because between the SUV and me was my wheelchair, which was hit first by the impact. My left leg was crushed and I lay in bed in great pain for weeks. My accident in November last year already had serious consequences. At that time, I fell down a high curb that was not well marked. Also, a life-threatening situation that “fortunately only” cost me the hearing in my right ear. My body is not so robust and resilient, like a body without a disability. And the additional ailments are slowly but surely piling up. They weaken me and wear me down.

**My social value is linked to my performance and my functioning**.

The other part of the equation is my mind, and part of that is my internalized ableism. As a disabled person, I have internalized that my social value is linked to my performance and my functioning. Disabilities are looked at as if they were something bad, as if we people with disabilities were a financial and emotional burden for the family, environment, and society. Even more we try to gain our raison d’être by performing, doing things on our own. We often function beyond our own limits to keep up with able-bodied people. We feel that we are constantly being put to the test and feel it is our duty to make our disability forget. This is physically and mentally exhausting for us and gives us another task.

In combination, these two aspects are downright fatal. Because the less my body can do, the heavier the ableistic pressure on my shoulders to function. I try ever more desperately to make up for my shortcomings and ever more resolutely ignore all warning signs as I march beyond my own limits.

I often wonder what to do. I am an ambitious person who likes to set goals. They used to inspire me, now I’m afraid of them. Because I know myself and I know the ableism in me.
When I have a goal, I work even harder to achieve it. But I know deep down that the goal I’m approaching is not the result of my project. The word BURNOUT is written in big letters on the black and white tiled banner slowly but surely coming my way.

**SO, MY HEALTH ISSUES, COUPLED WITH A TENDENCY TO OVERPLAY MY DIAGNOSES, COME AT A PRICE**

In the article *Why People Hide Their Disabilities in the Workplace*, the authors of the Harvard Business Review brilliantly summarized the problem of burnout caused by hiding disabilities with the following introduction:

“If you hide a disability, everyday life (...) becomes a much heavier burden. They live in fear of being discovered. You work overtime to hide the real you.”

**Necessary expenses quickly add up, and can lead to additional worries**

People who hide their disability are two times more likely to be anxious and four times more likely to feel isolated. This contributes to overload and can tip the scales of burnout. There are also additional financial burdens. Many things such as PAs or mobility aids have to be paid for by disabled people themselves or at high cost. Even barrier-free living space is often hardly affordable. These hidden extra costs that a disability entails are known in the community as "crip tax", "crip costs" or "crip time". The necessary expenses quickly add up and can lead to additional worries. Especially when you live in constant fear of losing your job and no longer being able to afford everyday utensils.

**Society inherently prejudices that we can hardly perform or do a good job.**

It's a game of hide and seek with no winner. You've built up the illusion of a level of performance that you can't maintain without physical and/or mental cost. But it's hard to backtrack, to uncover the masquerade and explain that in reality you have a disability, you've worked beyond your limits and now you can't afford as much. Ableism is brutal and painful to endure. Society expects very little from people with disabilities. It inherently prejudices that we can hardly perform or do a good job. Of course, there is a great incentive for a disabled employee to refute these prejudices. You're trying to prove to yourself that you're good, is strong and energetic and makes a valuable contribution to the company. And if you then cave in and supposedly confirm the negative prejudice, it's a devastating admission. Not only for self-respect, but also for reasons of solidarity with other disabled people. What if the company doesn't hire more people with disabilities now in the future because I confirmed the negative image I was trying to disprove?

**THE BURDEN ON THE SHOULDERS OF DISABLED WORKERS IS ENORMOUS**

I would go so far as to say that every able-bodied person employed anywhere with a disability at the back of their minds has the fear of losing a good job because their condition deteriorates. The mess gets even worse when people with disabilities work part-time for low pay or are employed in workshops for disabled people. Being stuck in a job that doesn't see you and in which you feel trapped with no alternatives and no opportunities to learn or advance can also create stress, which in turn can lead to burnout.
Constantly "asking for help" is also corrosive and can contribute to burnout. I find that I ask less for help when I'm not feeling well. Ultimately, however, this makes me feel even more helpless and my self-confidence drops, which in turn can lead to a flare-up of my imposter syndrome: a feeling of massive self-doubt about one's own performance. This "reliance burnout" can cause people with disabilities to feel that they are unable to cope with the demands of their jobs or daily life activities.

**THE WAY OUT OF THIS DILEMMA IS ACTUALLY CLEAR: CONSISTENT ANTI-ABLEISM**

But it is heavy. It means radical acceptance of what is - physically and mentally. Thus, it forces you to see, draw and hold your own limits. It's about protecting yourself from the ableist idea that self-worth is tied to achievement. And it demands openness – language. For a large part of internalized ableism is shame. And shame grows in the unspoken, and only goes away when we find words for it. It's hard work, but it's definitely worth it.

Because as long as I go full throttle, I never find that I can reach my destination at a slower pace. And as long as I'm fighting the reality that I need help, asking for something will be a struggle every time.

I think a first step is to heed an old adage: "The perfect is the enemy of the good." (Voltaire)

By not recognizing the good as enough and always wanting to achieve more, I burn out in the sprint in the last few meters between good and perfect.

I'd like to find out what happens when I stop wasting all my energy working off my internalized ableism. It's annoying. It wants to make me believe that I'm only worth something if I achieve things or manage them on my own. I don't have the answer yet. But I guess I would then have more time and space for things that not only draw energy, but also give it back.

And there are a few things that would come to mind: Spend more time with friends or my partner, eat well, sleep well, watch a movie, go for walks. Wouldn't it be nice to find out?
Who are you and what is your relationship to OI?
My name is Nick Bishop and I am professor of paediatric bone diseases at the University of Sheffield in the UK. I’ve worked here for the last 25 years (nearly). When I first came to Sheffield, I set up a metabolic bone disease unit for children, which was the first such unit to be purely bone orientated in the UK. Most of the children that I looked after then, and over the following years, had OI. so I guess you could say that most of my professional life has been looking after children and families with osteogenesis imperfecta and undertaking research to try and improve outcomes for them

You recently retired - is this a real retirement or are you continuing to work in secret?
I am “mostly retired”! I spend around one day per week working on the REMEDI4ALL demonstrator project, which is looking at the use of losartan, a medicine normally used to treat high blood pressure, in adults and older adolescents with osteogenesis imperfecta.

Tell us about the Remedi4All project!
The project that I am working on is one of four “demonstrator projects” run by the REMEDI4ALL consortium. The overall consortium was funded by a large grant from the European Union – €25 million and the coordinating group was developed from another European consortium called EATRIS. The main people steering, the consortium are Anton Ussi, Don Lo and Martin de Kort. The whole purpose of the consortium is to create a system which can look at medicines that already exist but could be used for different conditions. So there are a lot of “work packages”, including one that focuses on the voice and needs of patients forming the consortium group. The demonstrator projects are all in a single work package.

For our specific demonstrator project, we are using centres in the UK and Italy only and to recruit 30 patients aged 16 and over to take losartan for 24 weeks. We are looking to find out whether using losartan reduces the speed with which bone is destroyed and then remade, which is normally increased in osteogenesis imperfecta.

Tell us about your career! How did it all start?
When and how did you start working with OI?
I started off as a paediatrician interested in tiny babies and spent 10 years doing clinical practice and researching that area focusing on those born both small and early. As part of the original research, I did in that area, we needed to understand how the skeleton developed after that early life period and so I started measuring bone density in apparently normal children!
After a while, I started getting letters and calls asking if I could help with children who had got problems with bones, particularly fragile bones. I realised that I knew virtually nothing about bone disease in older children and ended up getting a fellowship to go to work with Francis Glorieux in Montréal for two years. That’s where I really learnt about osteogenesis imperfecta and brought that knowledge back to the UK.

Have you worked most as a clinician or researcher?
Since the time when I worked with premature babies, my effort has been spent equally working on clinical delivery and research to try and improve outcome, so I would say half and half!

Are there happenings that have made a special impression on you during your career?
I think there have been so many, that it will be difficult to pick out just one or two; every child with OI and every family has their own story, and I hope each one feels that they have been looked after as individuals by us in Sheffield. I think the time we spent as a family in Montréal was memorable, not just because of the clinical work, but because we made a huge number of friends who we go back to see on a regular basis.

Are there things in our career you regret?
Certainly; there are particular moments when I could have done a lot better but I think any doctor would tell you that. Overall, I guess my biggest regret is not being able to get the losartan study done in children, because we lost access to the liquid form of losartan which we wanted to use, because the manufacturer stopped producing it, just as the study was about to start.

What are most proud of during your career?
This might seem out of place in talking about OI, but I think the piece of research I’ve done that has made the biggest impact was the very first one in premature babies where we showed that aluminium
contaminating their intravenous feeding solutions was causing brain damage. We managed to get the aluminium content of those feeding solutions lowered in legislation both in Europe and the United States as a result of the work that we did. This will have had a major effect on the neurodevelopment of thousands of premature babies in many countries over the last 20 years, so I am very proud of that.

Other highlights?
Setting up the paediatric metabolic bone disease clinic in Sheffield and growing and developing the team there; getting the highly specialised service for children with OI established in England with four centres providing multidisciplinary care; doing the study with risendronate, that showed a positive effect on reducing fractures in children with mild OI; being the first Centre in Europe to use the enzyme replacement therapy asfotase alfa for a child with hypophosphatasia; being the director of the first clinical research facility in a Children’s Hospital in the UK; my time as vice president for science and research at the Royal College of paediatrics and child health during the Covid pandemic which was pretty intense.

After OI2022 I felt a bit like a balloon after a party; it was such an enjoyable meeting for me, seeing so many friends and colleagues, all together in one place as the pandemic released its hold on us all - and then it all stopped!

During the years you have developed a close relationship to people with OI and the OI-community, especially the BBS. How has this influenced your way of thinking about OI?
I have always tried in my clinical work, as well as my research, to see things from the point of view of those who are “on the receiving end” - both the children and their families; I think being able to work with organisations, such as the BBS, has simply reinforced that.

You have always been a champion for more patient involvement (PPI). Why is it important?
For me, PPI means listening to what patients want and making sure that it’s reflected in what I do whether that is clinical work or research. So, for instance, in the research study, we are doing, we have had patient input right from the very beginning in terms of the design of the study and thinking about what we can reasonably ask people to do. I expect that that involvement will continue throughout the course of the study, and in fact I would regard the ideal situation as being one of “co-production”. PPI is vital for research, because otherwise you lose that essential perspective which makes the study relevant and important for the OI individual.
Organisations can support in so many different ways - from directly contributing to the running of a study, to ensuring that the membership are aware of what is happening and can participate as individuals in things like trial steering committees and expert patient groups. Patient organisations can also act as a vital source of information for patients about studies and disseminate information from ongoing studies. This helps people understand what’s going on and encourages greater participation, whether or not that leads to actually being directly involved in the study itself.

**What are the major things that have changed/developed in OI-research in the last 20 years?**
I like to think that in the paediatric area we have led the way in trying to develop multidisciplinary services that are properly integrated for the care of children and their families. To me this has been the biggest advance in the last 20 years alongside the use of bisphosphonates. We are now in a period when new medicines may become available and that’s really exciting, but I will have to leave that to others to implement now I have retired.

**Have most of the knowledge gaps in OI now been filled?**
I don’t think most of the knowledge gaps have been filled yet. OI is such a complicated condition affecting not just the bones, but the muscles, lungs, heart and other tissues as well. We have a better understanding now of what is happening in the bones, but there is still plenty to find out about what is happening elsewhere in the body.

**We’re living in exciting times - there have never been so many new treatments and therapies being investigated for OI as now. What are your thoughts on this? Are you optimistic we will find "the cure" or a more efficient therapy to alleviate symptoms?**
Perhaps one day with gene therapy, but until we can correct the fundamental defect that produces abnormal structural proteins, like type one collagen - which is the problem for most people who have OI - any treatment will not be a cure, but simply something that treats the consequences of the underlying genetic problem. How far away is gene therapy? Difficult to answer because we’ve been promised that it’s coming for almost all the time I have been in clinical practice - so I suspect that it will be another 10 to 15 years at least.
How do we deal with death among our members?

By Ute Wallentin, OIFE Social Network and Board member of the German OI-Association (DOIG)

When OI-children or adults or members in general from our association die, how do we want to deal with the news of their death? And if they die (much too) young, what offers can we make to the surviving relatives? Ute Wallentin has made some thoughts about this and collected suggestions.

For a long time I brooded over this text and weighed whether and how to write it, and why now. At the end of January, winter had broken in again, with lots of snow and "dangerous" ice. Everything suddenly seemed even colder, empty and dead. But we all know from experience that buds will soon grow under the snow on the trees, that life goes on and soon everything will be green again.

Still, sometimes I feel sad because a year ago our OI mom died and in January a dear, young and energetic OI friend died all of a sudden and left everyone incredulous and shocked.

It is not always easy for someone with OI to reach an advanced age

In the past year, the death of my OI-mother, who was 86 years old and full of life, saddened many people as much as it saddened us. But on the other hand, within my large and international OI family, I have also been able to give rather great encouragement with this sad news to quite a few others, because it is not always easy for someone with OI, damaged heart valves, a lot of pain and rotten connective tissue, to reach such an advanced age. But fortunately many of us succeed! On the other hand, the fact that another very young man, only 34 years old, just had to die of a stupid pneumonia is and remains tragic and we are inconsolable and sad about it.

How do we deal with deaths among our members?

Especially this sad loss raises again an important question in our German association DOIG which some of us are always concerned about: how do we deal with deaths among our members? Why do we usually hardly inform our members about it and how do we try to help the relatives and friends of our deceased through the then following hard times, to accompany them somehow despite our own helplessness?

When I think about this, or when I talk to others, we come up with quite a large number of families who have had to say goodbye to their children. These deaths were not always related to OI, even perfectly healthy, non-disabled young people die sometimes far too early. But in many cases a connection with OI was recognizable and this can of course cause fear, for OI patients themselves and for their parents and relatives. That is why we have been so extremely cautious about this topic so far. But that is about to change. We are looking for a form that does not trigger fear, but shows the bereaved that they are not forgotten, that we are very happy if they continue to be members and stay connected to us!

Some of them, unfortunately only a few so far, even continue to come regularly to our events, like the older couple who continue to come to adult seminars and often to the annual meeting. Like others who after losing their OI baby, still actively participate and offer workshops at our conferences. Or like Angelika and family, who year after year and lovingly takes care of our toddlers in the nursery at the annual meeting in Duderstadt, even though she lost her first daughter in infancy. Hats off, to all these and thank you!
But what about all those who remain members after such a death, but don’t have the heart to come on their own when we meet? They, too, continue to be welcome and we talk among ourselves again and again about these families as well as about the great people we have all lost but not forgotten. But unfortunately, the bereaved relatives hardly notice and know that. Are we doing this out of shyness about death and grief, out of respect for their loss? But couldn’t we do it differently and better?

Years ago, we decided against printing obituaries in our OI-journal “Durchbruch” (=breakthrough) so as not to frighten anyone. I still think that’s okay, in a way. But unfortunately, we have also left the relatives perhaps too much alone with their grief.

Recently, after about 20 years, I finally met an orphaned OI father again, whose daughter had died at a very young age. I know that he very courageously faced his grief and trained as a grief counselor and is still actively dealing with it today. This impresses me very much and gave me the idea that within our association we should please deal with this difficult and painful topic in a different way.

**Virtual commemoration on myDOIG**
Within the protected framework of our members’ forum meineDOIG we could commemorate in an appropriate way - and of course only with the consent of the relatives - those who are unfortunately no longer physically and tangibly among us, but whom we do not want to forget. Like our two former chairmen Peter Radtke and Kall Henn, but also many others who were not so well known. On Facebook we still see photos of some of them from time to time, which always reminds us that they could no longer post them themselves. Around the campfire in Duderstadt at our annual conference or in conversation with each other, we tell each other funny and thoughtful stories and can even laugh together about some memories. And that feels so good!

**Mediation of mourning groups, own counseling offer**
We might have the possibility, for example through our membership in the umbrella organization we belong to “Bundesverband Kinderhospiz” (childrens hospice), to arrange contacts to mourning groups. But it would also be possible to establish another internal counselling offer within our association, because a psychotherapist who is herself married to one of our adult members would be happy to accompany individual members or possibly groups on this topic. This would be possible, similar to the comprehensive counselling offer we have had for two years already via telephone or Zoom.

**Please get in touch!**
We ask very cordially for reactions of our readers to these considerations! I would be pleased to hear or read from those who feel addressed here and to hear your opinion. All reactions and thoughts are very welcome, not only from inside Germany or Europe!

Best regards!
Ute
OI & NUTRITION
Calcium is an essential ingredient for good bone health, which is also very important for people with OI. But which foods have the most calcium? Find out by downloading this list of the approximate calcium content of common foods, available in multiple languages: https://bit.ly/44MkIsn

OI PANAMA
The Panama OI-community has a newly reestablished OI-organization called Familias OI Panamá run by and for people with OI themselves. You can follow them on social media:
Facebook: https://www.facebook.com/FOIPanama
Instagram: https://www.instagram.com/p/CvkYv5jOw5S/

LEONARDO ON AIR
Listen to the newRare on Air episode from EURORDIS where Leonardo Panzeri from the Italian OI-association ASITOI explains why repurposing of existing drugs, can be a smart strategy for rare conditions like OI. Find the podcast here: www.eurordis.org/rare-on-air/

EXPLORING TOGETHER – ASITOI
It was a very nice weekend with the project "EsplorandoInsieme" (Exploring Together) which saw children and their families visiting "Le Cornelle" Zoo and also included an experience with the New Pop Orchestra. This is part of a project called "Growing up together" organized by ASITOI where OI-children and their families are involved.
NEW OIFE PHOTO GALLERY
Did you know that OIFE's webpage now has a photo gallery, where we will post photos from our major events? We have published all the photos from the Stockholm conference there and you can also find photos from our topical meeting in Riga, Latvia: https://oife.org/news-resources/photo-gallery/

ADVOCACY FOR OI
In June the BBS (UK & Ireland) hosted an event at the British Parliament, with the purpose of advocating for better services for children and adults with OI and other rare bone conditions.

GREETINGS FROM THE NETHERLANDS
Every year the Dutch organization VOI have their annual swim day. On June 24th they were 52 people who met and had a good time together. Also new members, which is great!

GENETICS FOR YOUTH
The RARE Youth Revolution "My life, My genetics project" is out now! It is an informative series on genetics and relationships filmed by young people for young people. Follow six young individuals as they explore the following topics: genetic inheritance, family planning, genetic counselling, newborn screening and prenatal screening, including a tour of the Illumina labs. Find out more here: https://bit.ly/43OGT01

GREETINGS FROM GERMANY
The German Annual Meeting took place the same week-end as the Stockholm conference. See some glimpses below.

SWEDISH OI-INFORMATION
The Swedish health authorities for rare conditions have updated their national information about osteogenesis imperfecta. https://tinyurl.com/32knef4h
Research Announcements

Sanofi POISE1
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.

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 we will consider expanding enrollment criteria in future studies.

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 roles 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.

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.

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 under ClinicalTrials.gov NCT05231668.

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 

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. The study is focusing on reduction in fracture rate, including morphometric vertebral fractures, in younger pediatric 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.

For more information on both of these trials, please reach out to trialrecruitment@ultragenyx.com or OIStudyInfo@ultragenyx.com.
OIFE Investigator Meeting – Registration & Abstract Submission open

OIFE together with our Medical Advisory Board, would like to invite you to register for the second virtual European Investigator Meeting for osteogenesis imperfecta (OI).

**TARGET GROUP**
- Primarily researchers and clinicians working with OI in Europe and other countries, and will not be adapted to a lay audience.
- Patient representatives from OI organizations can attend if they have a special interest in research and development.
- Industry representatives are also welcome to attend.

**PROGRAMME**
The programme will include presentations and discussion on the following topics:
- Research collaboration in Europe: hurdles and opportunities
- What’s new in basic science?
- Methods and tools to evaluate outcomes in clinical trials
- Patient involvement in research
- Nosology and classification in OI

Link to register: [https://forms.gle/m3iS1Y2x4m5MF4y58](https://forms.gle/m3iS1Y2x4m5MF4y58)

Abstract submission is open
A number of oral slots are available on the programme for submitted abstracts on OI research. Priority will be given to abstracts describing novel research and hot topics. We invite your abstracts on any aspect of OI research, whether basic, translational or clinical. Deadline for abstracts: Monday 9 October 23:59 CET.

[Abstract submission form](https://forms.gle/m3iS1Y2x4m5MF4y58)  [Abstract submission guidelines](https://oife.org/what-we-do/events/oife-investigator-meeting-2023/)

More information can be found here: [https://oife.org/what-we-do/events/oife-investigator-meeting-2023/](https://oife.org/what-we-do/events/oife-investigator-meeting-2023/)
Webinar - OIFE Update on Clinical Trials

We invite you to register for a 1.5 hour webinar called "Clinical Trial Update 2023". The goal of the webinar is to update the OI-community about ongoing clinical trials - both those who are recruiting and others.

This includes the following trials:
- The Cosmic and the Orbit trials (setrsumab)
- The BoostB4 trial (stem cells)
- The Topaz trial (teriparatide & zolendronate)
- The Poise 1 study (SAR439459)

Link to register (for free): https://forms.gle/krsjjo8RnvV2b6sj8

TARGET GROUP
The target group is anyone interested in OI-related research - both professionals and people with OI and their families can attend.

REGISTRATION DEADLINE
October 20th 2023 23.59 CET

QUESTIONS?
Please send any questions you might have about the event to office@oife.org

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