OIFE and its member organizations were well represented at the ERN BOND meeting in Bologna, Italy in May.
From the left: Rebecca, Inger-Margrethe, Claudia and Leonardo.

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Editorial

By Malene Sillas, OIFE Board member

GROW UP AND SPEAK UP!

Growing up from childhood, becoming a young adult, I’ve always felt that whatever I do, it feels like I’m the first person to do it. I remember that the first time I felt that way, I was in my second year of high school. The whole class was going on a school trip which had been hyped up for us since we started our first year. The class was gathered, and our teachers were presenting to us, which country we were going to. It ended up being Malta, and we were all ecstatic – going to a new country with all our best friends. Besides seeing churches and museums with our teachers, most of us just looked forward to the partying.

After we were told the news, the teachers pulled me aside, they told me, that it wasn’t sure that I was able to come, because the accommodation that was the cheapest had no elevator and loads of stairs. I remember just feeling this huge sense of unfairness. That was my first time speaking up for my rights, and after threatening them with the UN’s convention on the Rights of Persons with Disabilities, I was able to go.
I’ve since then wondered how much easier my classmates and peers have had it growing up. Because besides the normal struggles and worries about fitting in, getting good grades, and making friends, there was just this huge mountain of questions and worries I had to overcome besides all normal teenage-young adult worries. I’m not one to complain, but my point is rather that growing up with OI, you have to learn to speak up besides learning how to drive, how to solve complicated math problems, and how to fit in. It’s just as important.

You have to become an expert in navigating the system, talking to doctors (which is a whole story in itself), and so much more that none of your ‘able-bodied’ friends have ever even considered as a problem. To be honest, I think my parents gave me the greatest gift of all by becoming a member of the Danish OI society DFOI when I was a baby. Fortunate for me, we were a large group of girls born with OI within a few years. And even today it means everything to have these friends to talk to about the small struggles and worries that come with this OI-life. My OI friends fully understand me and have good inputs on how they handled similar situations.

This is the core of why I believe that OI-organizations are the most important thing that ever happened to me. I think that people who decide not to participate in national OI-meetings, miss out on so much, even if they only have a very mild OI. Because no matter what, you’re going to face challenges that nobody besides people with OI knows about.

To me, speaking up about things that were unfair, was something I over the years came to terms with. Mostly because I started to face them early on. But what came as a surprise for me, was the fact that I wasn’t ‘done’ with learning about how to speak up.

A few years ago, I noticed that the thing that I was struggling with, and had been for some time, was speaking up about my challenges and worries to my ‘able-bodied’ friends. For a long time, I was super conscious about talking to my ‘able-bodied’ friends about these struggles, because my worst fear was, that they would look at me as their complaining disabled-friend and not as me. And it even got so bad that I lied about what I was doing on a weekend if I was attending an OI-related event. I didn’t want to become my disability. I remember quite vividly how I was telling my friends how I was going to a boring conference, when in reality, I was going to a youth event. This is ridiculous, especially if you have ever attended an OIFE youth event. Then you know that it’s not a boring conference. I’ve since been working on this. But it made me realize that learning to speak up is never something I’ve finished learning – it’s an ongoing journey.

Malene Sillas, OIFE Board member

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

Between February and June we have been focusing on preparing our big conference in June, the OIFE Youth Event and being represented in a number of other arenas. We also had the chance to gather the new OIFE Board in Stockholm – to discuss future strategies, but also to test the conference hotel Scandic Continental. We also held two OIFE Drop In sessions, for the OIFE representatives and a social After Party event for Wishbone Day.
MEETINGS & WEBINARS
In addition to internal meetings, and meetings with our members, we have had large number of meetings connected to the Stockholm conference. We have also had external meetings with our normal collaborators incl. the XLH Alliance and some meetings connected to different clinical trials. This is a list of the most important meetings we have attended in addition:

- OIFE Board meetings Feb 21, March 21, April 18 and April 28-30 face to face.
- EJP RD application – Natural History Projects, Feb 8 (IW + Antonella and Oliver from MAB)
- European Hemophilia Consortium Think Tank on care pathways, Feb 14 (IW)
- EURORDIS Alumni - how to apply to a call for research projects Feb 23 (IW)
- Steering Committee IMPACT survey Feb 24 (IW and TvW)
- Researcher Jessica Eaton (child abuse project), Feb 27 (IW)
- Adult Health Initiatives, Feb 28th, March 9, April 20 (IW)
- Innoskel meeting about PROMs and endpoints, March 1 (IW)
- TeleECHO Bone Turnover Markers, March 2 (IW)
- ICD11 & OI potential project (IW and IMSP)
- Rare Bone Disease Alliance Scientific Symposium on OI online, March 9-10 (IW and ISMP)
- GlobeReg March 10 (IW)
- Intro meeting Metabolic support UK March 14 (IW)

EuRR-Bone Meetings
On February 13th we were four representatives from OIFE who attended the final meeting of the EuRR-Bone registry as a 3 year old project. Claudia Finis attended face to face (see photo) and Taco van Welzenis, Rebecca Tvedt Skarberg and Ingunn Westerheim attended online. The project team in Leiden have delivered on all their milestones, and has achieved a lot in these 3 years. However – the awareness about the registry is still too low and too few centers in Europe are contributing with data – especially to the CORE registry. So there are plenty of challenges to solve in the next 3 years, including securing more predictable funding. OIFE will continue to be actively involved in the groups for the disease specific modules (vertical themes), which are now called study groups.

On Monday April 3rd the first joint EuRECa / EuRR-Bone symposium took place at the Academy building in Leiden, the Netherlands. Claudia Finis attended face to face and Ingunn and others attended online. In addition to an overview from the perspective of the ERNs, the professionals and the patients, there were also presentations from the disease specific modules – including the one for OI. Faisal
Ahmed talked about lessons learned and Natasha Appelmann-Dijkstra presented future perspectives. You can watch the recording of the meeting here.

EuRREB, the European Registries for Rare Endocrine and Bone Conditions consists of the EuRRECa and EuRR-Bone project. The aim is to be Europe’s most comprehensive and accessible resource for researchers studying rare endocrine and bone/mineral conditions, supported by both Endo-ERN and ERN BOND, scientific and patient societies. The new URL is https://eurreb.eu/

**Pain and OI Project & Pain and OI survey**

Together with the OI Foundation, OIFE organized the 6th workshop in the Pain & OI project on March 27. Ingunn and Ute Wallentin represented the OIFE. Kelly Thorstad gave an introduction to educational resources from the Shriners Montreal, Ariane Kwiet and Ingunn presented the plans for the Pain and OI Toolbox and Michael Stewart presented the preliminary results from the pain and OI survey, which researchers from Baylor College of Medicine will help us analyze. Initial findings include:

1. Many people with OI appear to experience lots of chronic pain.
2. Back pain is common for people who completed the survey.
3. Pain impacts many parts of people’s lives including career and social life decisions.

In May 2022 OIFE and the OIF agreed to be part of a steering committee on Sanofi’s pain project which includes a literature search and qualitative interviews in 4-5 different countries, incl. UK, US and Australia. The goal is to investigate how and if pain can be used as potential outcome measure in clinical trials. In addition to Tracy Hart from the OIF, Ingunn Westerheim and Ute Wallentin has represented OIFE in meetings on March 3, March 6 and April 21.

**ADVANCED THERAPIES SEMINAR**

The snowfall didn’t stop OI from being very well represented at a very educational seminar on challenges and possibilities in advanced therapies March 13. The event was hosted by Oslo University Hospital in Norway. Rebecca Tvedt Skarberg was in the organizing committee, Inger-Margrethe from NFOI gave a talk about the importance of patient involvement throughout the process and Ingunn from OIFE was among the many interested participants. Advanced therapies (ATMPs) include therapies like gene therapies and stem cell treatments.

**OIFE AT NORDIC RARE DISEASE SUMMIT**

On April 17 OIFE’s president Ingunn was invited to talk at the Nordic Rare Disease Summit (hybrid), which was attended by more than 600 participants - 50% live and 50% virtually. Ingunn attended online and took part as a panelist in the session called Access to innovation - about how people with rare conditions in Europe can have quicker access to new and innovative therapies. There were also other familiar faces to be seen from the OI- and Rare Disease-community.
OIFE IN BOLOGNA
The OIFE was well represented at the spring meeting of ERN BOND in Bologna the first week of May. ERN BOND brings together 53 health care providers (hospitals) that deal with rare bone conditions. 40 people attended face to face and 13 online. In addition to several members of the OIFE MAB, Rebecca and Claudia represented OIFE and Inger-Margrethe and Leonardo from the Norwegian and Italian OI-organizations were there. Future plans and priorities in the European Reference Network were discussed. On May 6th the seminar "Patient priorities in ERN BOND beyond Quality of Life" took place, organized by the patient representatives in ERN BOND. The aim was to shine a light on four priority areas, identified by in a dialogue between patients and clinicians. Topics included pregnancy, movement analysis and mobility, transition and pain.

PATIENT INVOLVEMENT IN PRECISION MEDICINE
From May 4-5 Ingunn attended a large Norwegian conference about precision medicine (personalized medicine) – which included many different talks about how innovative research and strategies can help develop new and more precise treatments for cancer, rare conditions, diabetes, depression and more. Both researchers, clinicians, patient representatives and policy makers were present. Together with Ann-Rita from the Norwegian Cancer Society, Ingunn gave a 30 minute talk about patient involvement – on an individual level, on a policy level and in research and care management.

EURORDIS AGM & OIFE MEMBERSHIP MEETINGS
Malene Sillas represented OIFE at the EURORDIS AGM (online) on May 17th, where a new board was elected, annual reports approved and new strategies discussed. Lars Romundstad will represent OIFE at the EURORDIS Membership Meeting in Stockholm from May 25-27. Inger-Margrethe (NFOI) and Rebecca Tvedt Skarberg (EURORDIS Board member) will also be there to represent the OI-community.
Meet the new OIFE 2\textsuperscript{nd} Youth Coordinator

MATILDE MEDUM NIELSEN, DENMARK

I’m Matilde, from Denmark, and I was born with OI type III. I was raised and have been living in Aarhus my entire life until July 2022, where I moved to Townsville in Australia to fulfill my dream of becoming a marine biologist. Here I study a master’s in marine biology, which I absolutely love! Besides my studies I also enjoy swimming, kayak paddling, and most recently scuba diving.

I think one of the main reasons I love the ocean, is because I feel so free, when I’m in the water. There’s no pain and no limits or restrictions since you feel almost weightless in water. In the ocean there is also relatively few obstacles you can accidentally injure yourself on and so the risk of a fracture decreases. As people with OI, I’m sure we all value those moments where we’re able to feel bodily freedom, because we don’t have to worry too much about the possibility of a broken bone or our muscles aching. At least that is how I feel, when I’m in the water.

By becoming a part of the OIFE Youth coordinator team, I hope to be the support, that some young people with OI might need, whether it is regarding health, social life, education, work, or something else. I also hope I can inspire young people with OI, to fulfill their own dreams and achieve what they desire in their life, by sharing my own thoughts and experiences.

Besides providing support, it is also part of my task as a OIFE coordinator to organize the yearly Youth Events. I have participated in 6 events myself, and I would highly recommend all young OI’s to participate in an event at some point. It is a great opportunity to bond with other young people with OI from different countries and share experiences and stories. Here I have found some of my very best friends, that knows exactly how it is to live a life with OI. I think it is very important to be met with comprehension for the struggles we all deal with - but also just to have a lot of fun together.

Feel free to always get in touch with me if you have any questions or need support (email: matildemn@hotmail.com or Facebook: https://www.facebook.com/matilde.medumnielsen), and I will do my very best to get back to you as soon as possible!
The IMPACT Survey - how we identified knowledge gaps and created a meaningful survey for the OI-community

The IMPACT Survey was an international research project exploring the real impact OI has on people’s lives. The survey was run by Sam and Maria from the Wickenstones Company - an experienced scientific agency who, OIFE, OIF and Mereo Biopharma have engaged to do the work. Five articles will be written with the support of Wickenstones and the first one about the initial literature search and scoping review has already been published in Orphanet journal.

Who are you & what is your relationship to OI?

Sam:
I am a market access consultant, working with pharmaceutical companies to help bring new medicines and interventions to patients with all types of conditions. Over the last 3 years, I have been heading up the IMPACT Survey, and had the great opportunity to learn a lot about OI and to working closely with OI clinical experts and community members.

Maria:
I am an employee at a market access agency - so I came to OI completely by accident. A few years ago, I started a new job and the IMPACT Survey was one of the first projects I worked on. Before working on IMPACT and with the patient community I knew very little about the condition OI, but now this work has become my favourite task at work.

What is your professional background?

Maria: I have a Master’s degree in Immunology. After finishing my degree, I wanted to work outside of academia but stay close to scientific work, so I started to work as a scientific writer. It felt like the best of both worlds – reading lots of scientific papers, while having more opportunities than academia might offer.
Sam: I have a PhD in the field of medical and molecular genetics from the University of Birmingham where I was conducting focussed research on a very rare inherited condition called Wolfram syndrome. I am fascinated by biology and medicine, particularly rare genetic conditions. Back in the lab, I got to work on the basics of understanding how diseases come about. These days, I now work to apply my background knowledge and interests in a field where I can contribute to people getting access to new and innovative treatments, which is very rewarding.

What does a company like Wickenstones do?
Wickenstones is a global ‘market access’ consultancy, which means we help clients from all around the globe bring new medicines to patients who need them. Our work involves a whole spectrum of things, including planning strategies for rolling out new treatments, gathering data and evidence to understand whether new drugs will meet an existing need, helping to communicate the value of medicines to the people who make decisions about their availability, and looking at ways of getting around specific barriers to patient access in different countries, whether those be economic, clinical or policy-related challenges.

Tell us about the background for the scoping review!
When we started to plan the IMPACT Survey it very quickly became clear that we needed to understand what was known about the condition OI and the OI community. To make IMPACT as useful as possible we wanted to know where the gaps were in the published scientific literature. IMPACT was designed based on the findings of the review and includes many questions we would have never thought to ask if we had not done the review first. The review was systematic – which means that we set out stringent criteria for the search and selection of relevant publications to make sure to capture everything.

Did you search in grey literature - what is it?
Grey literature is any literature that isn’t published in peer reviewed scientific journals. Peer review means when scientists submit their research for publication, other experts in the field will review their submission to make sure that it is sufficiently high quality. So grey literature is basically everything else – for example information from patient organisations, healthcare providers and charities. While such information cannot replace scientific publications, it can help scientists to understand what patients are actually concerned about and inform future research, as it did in this project.
How was it financed?
The review and the survey were both financed by Mereo BioPharma, a pharmaceutical company developing a new treatment for OI.

How did you use the scoping review results when creating the survey?
When we finished the review, we made a list of all the gaps we had found in the research. For some topics we had only found very few datapoints, or only insights from interviews with patients. For others, only information on some patient groups but not others, was available (for example only research in children but not adults). We discussed the gaps with everyone involved in the project, OI researchers and patients, to understand which ones were most relevant. Then we considered which topics were most suitable to research in an online survey. Not every question can be answered through survey data, but many can be. For example, questions about the experience of people with OI and their families can be answered in this way.

What are the challenges of doing a good literature search? How did you choose the right keywords?
The searches are actually more straightforward than one would think. Scientific literature is organised in databases that have good tools for searching and with some experience and trial and error, one can be pretty certain to find the right information. The real challenge with this work was to make sense of our search results. We had over 200 reports to read and sort into clear categories to make sense of what information they contained. This can be quite tricky, because a lot of research in the field is based on interviews with patients, and while those interviews brought up tons of interesting information it was difficult to pull out information from such research and get an overview.

What were your most interesting findings?
Identifying the biggest knowledge gaps was the most interesting aspect of the review, particularly when we talked to experts and community members about the things they felt were most important to the medical and OI communities. This was invaluable for shaping the survey questions.

What are the most important knowledge gaps?
There’s a larger volume of research into bone health and the clinical aspects of OI compared to other topics. Significant data gaps exist around the socio-economic impact of OI on people with OI, their families, and wider society. There is also a lack of data on wider concerns beyond bone health, access and standards of OI healthcare and how health-related quality of life changes throughout life stages. There is also an underrepresentation of males with OI, young adults, and siblings of children with OI. Here you can read the open access article called “The patient clinical journey and socioeconomic impact of osteogenesis imperfecta: a systematic scoping review”.

Were there any surprising findings?
The lack of data on some of the things people say are very important, such as pain.

Why do you think we need socioeconomic data on OI?
Socioeconomic data serves to highlight the unmet needs of the OI community in terms of expenses incurred, access to services and care, and other financial burdens that people and their families may experience. Gathering this information enables decision-makers to better understand the value of interventions, policies, and healthcare access for the future.
What will come out of IMPACT in 2023 and 2024?

The high volume of responses means there are a lot of data to process. Over the coming year or two, the key findings of IMPACT from a global perspective will be submitted for publication in scientific journals and at some relevant congresses and meetings. The topics will cover the methods and demographics, economic impact, clinical and quality of life impact, and healthcare journey (care pathways) and experiences. Once the main findings have been shared, there will be many opportunities to look at the data from a country-specific perspective or focus down on certain questions or topics.

Did you expect as much as 2300 responses and has it created any challenges?

No, not at all! When we were conducting the literature review the biggest survey projects had a few hundred participants, so this is what we hoped we could achieve as well. When we hit 150 after a few days, I was stoked, even more so when we hit 500. But I did not expect the numbers to keep climbing the way they did. This has really only been possible because of the contribution of the OIFE and OIF, specifically Ingunn Westerheim, and Tracy Hart, who put so much effort into recruiting community members. The incredible response we got is of course a huge success – it means that the findings coming out for the survey will be more robust. It also allows us to analyse the data in more complex ways. However, it is also challenging. The survey is written to accommodate many different ages and respondent types which means that to be able to get an answer to a question, you first have find the relevant respondents within the database. Also the survey was available in 8 different languages, which is what enabled us to get this many respondents, but it of course also adds another layer of complexity, in both designing the survey and interpreting responses. For example, the same medicine may be called different things in different countries, and healthcare is organised in different ways. This means that when writing the survey (and now interpreting the results) we need to be mindful of that to make sure that our respondents understand the questions and we correctly interpret their answers.

In which ways did you have input from people with OI in the process?

We had input from people with OI throughout the entire process. There are members of the OI community on our project Steering Committee, who have provided invaluable insights and guidance to help us develop questions that are important and meaningful to people with OI and their families. We also had volunteers with OI from around the globe, who kindly gave up their time to review the survey in all 8 translations, help us to understand more about country-specific elements we may wish to consider, and to pilot test the electronic survey platform to make sure it worked. A whole network of community volunteers was also involved in survey roll out and recruitment, spreading the word and encouraging people to take part and have their say.

Any messages for the readers of OIFE Magazine?

Thank you! This project has been ground-breaking – the success of this initiative is down to the input, time and efforts of the community members who got involved.
Sweet home Chicago! – News from the OIF Scientific Meeting

By Dr. Lars Folkestad, OIFE Medical Advisory Board

Every year the OI Foundation (OIF) host a Scientific Meeting where researchers and clinicians from North America is invited for a 2-and-a-half-day long seminar focused on OI research. Lars Folkestad, member of OIFE’s Medical Advisory Board, joined the meeting in April and reports about it.

This year’s meeting was chaired by Ken Kozlof (left side of the photo) from the University of Michigan. As part of the collaboration between the OIF and the OIFE, I was invited to participate and present data from the Danish OI Register-Based Cohorts Studies that I have been working on for the better parts of 1.5 decades now. I would like to start by thanking both the OIF and OIFE for giving me the opportunity to participate.

I flew in Tuesday night and had a day to myself before the meeting. I took a walk in downtown Chicago and went on the ‘architectural boat ride’ – which takes on the Chicago River along many of the impressive high-rises and skyscrapers that make out the Chicago skyline. I am always impressed by these buildings, and it was nice to have a little time to see the big city.

The meeting started with a keynote presentation by Matthew Warman, who updated us on the newest animal model methods that are being used to produce OI mouse models with tissue specific phenotypes. In a near future, it will be possible to evaluate the respiratory system in a mouse, without having a mouse with severe skeletal problems in addition. This talk set the scene for what day 2 had in stall. Most of the talks covered the bone, cartilage, muscle cross talk and underlined again and again that you cannot have the one without the other. A talk that stood out for me, was the from Roy Morello, who has now produced a mouse model where you can turn on a specific OI mutation at any given time during the mouse lifetime. And you can do so in a tissue specific way. This means that you can have a mouse with healthy non-IOI bones, but OI muscle function – or a mouse with OI affected lungs, but no OI affected bones. The implication being that researchers will now be able to look at tissue specific outcomes without the doubt that we often have: Is what we see in this tissue related to OI related collagen changes or due to bone deformities that themselves give problems in other organs? One example of this dilemma would be lungs and scoliosis. Is the dyspnoea seen in some people with OI, related to changes in the lungs, or secondary to scoliosis and/or fractures to the spine and ribs? This new model (that do not have a name yet) will be able to differentiate, or in other words, look at OI related lung changes and OI bone related lung changes depending on where the researcher activates the mutation. For a clinician and clinical research like me who has never worked in a lab (sadly), this is nothing short of magic.
On the last day there were two sessions. The first updating us on the Brittle Bones Disorders Consortium (BBDC) studies – which is a large natural history study now including 1000 persons with OI from North America. The population have been followed in a systematic fashion, with a large body of data being collected. Just one example would be that the researchers have collected 4000 x-rays, mainly focused on fractures to the spine, prevalence, and severity of scoliosis. But also other bones have been evaluated. There are genetic screening and clinical evaluations of all participants. And looking at what studies the working group have planned, we can expect very interesting results from this study over the years to come. One key aspect of the study is to evaluate if a participant would fit into one of the many clinical treatment pathways that are set up using novel drugs to target different disease specific pathways in OI. These can be to improve bone health, reduce fracture risk and reduce pain. It was during this session that I gave my talk, and after a day of single cell studies I was a bit nervous that coming with population data would be a bit off topic. But the discussion was lively, and the questions had me on my toes. But it was fun, and I got some good points to follow-up on.

For me personally it was very special to be in a room with who’s-who of OI research in US and Canada, and I really enjoyed the discussions in the breaks, being invited out to eat after a long day of science and generally just bumping into people who had read my papers and who I have read many papers by. I really hope I will be invited back, because this was the best OI meeting I’ve been to since before COVID. Being able to actually shake hands with people is something that should not be disregarded as trivial. Yes, we can always Zoom! But sitting down at a table, talking about everything from stories about Dr. Raggio’s dog to growth curves for Canadian children with OI, and not worry about remembering to unmute yourself, just makes for a better conversation.

Surgery of adults with OI – interviews with specialized OI surgeons

In most countries, pediatric surgeons are not allowed to operate on adults with OI because of the way hospitals are organized. Usually, the surgery is performed by an adult physician who knows about one body part (like hip, fingers, knee, pelvis etc). What are the pros and cons of having OI-specialists, who can also operate adults? We asked three very experienced OI-surgeons for their opinion on this.

GUUS JANUS, ISALA ZWOLLE TREATMENT CENTER, THE NETHERLANDS

I’m Guus Janus (left side of the photo), orthopedic surgeon who have been working with people with OI since my internship. At the start I was astonished by the vulnerability of the bones, but also by the possibilities and impossibilities of surgical procedures in OI. As my interest in the disease and people grew, I started my PhD with the title “Osteogenesis Imperfecta. Orthopaedic and Fundamental aspects”.
I’m working at Isala Zwolle in the Netherlands, which is a large training hospital. Since I have been working quite a long time in Isala, I suppose I’ve operated more than hundreds of adult people with OI, from 18 years and older, with several complaints: Trauma, post-trauma abnormalities, deformity correction and arthritic joints. The last decade I preferably operate together with a colleague – orthopedic surgeon – to further improve quality. As often as possible, we implemented the same team for an operation of people with OI, to ensure the best possible care during and after the operation.

Do you sometimes refer to another adult specialist because of the type of surgery?
No. As an adult OI specialist I will accomplish all surgical procedures. An exception is foot surgery, a very experienced orthopedic foot-surgeon operates our patients, however sometimes with my assistance. All other procedures will be performed by me regularly and preferably together with one of my colleagues.

What are the pros and cons of having OI-specialists who can also operate adults?
An orthopedic surgeon with interest and experience in treating people with OI has an enormous added value. The decision process starts at the outpatient clinic and in consultation with the patient expectations about an operation are discussed. And it is precisely the knowledge and experience with OI that increases the quality of the procedure. Or sometimes it is wiser not to perform an intervention! An OI-specialist realizes the limitations and vulnerabilities of the bone and soft tissues in people with OI and can act accordingly.

Should we advocate for having more pediatric surgeons doing surgery on adults with OI?
I cannot judge about other health systems in other countries, but in the Netherlands the system with a very strict separation between children and adults works. Arjan Harsevoort, Nurse Practitioner and affiliated with our expertise center, attends two times a year the transition outpatient clinic at the WKZ, Utrecht, to coach the 17 and 18 years old ones to an adult expertise center.

Should it be a collaboration between a pediatric OI-surgeon and an adult specialized surgeon?
From my point of view also the adult benefits the most from a specialized orthopedic surgeon in OI and a dedicated team around the patient. I think OIFE and national patient organizations could play a role in this development, as well as pediatric OI-specialists.

Anything to add?
As an operation is serious business, it is not only the performing surgeon but also the OI-team of specialized doctors and paramedics which raises quality to a higher level. The ultimate success of a procedure is not only indication but also rehabilitation.

ROADMAP TO SURGERY

In 2021 the project Roadmap to Surgery came to an end. The purpose of the project was to create a roadmap for a standardized, integrated approach for optimal outcomes of surgery in OI, not only from a surgical view, but also from the patient’s perspective, that can be used in all infrastructures and cultures. The roadmap has been endorsed by the Study Group Genetics & Metabolic Diseases of the European Paediatric Orthopaedic Society and by the Executive Committee of the Osteogenesis Imperfecta Federation Europe and the Care4BrittleBones Foundation.

The article is open access and can be found here: https://www.tandfonline.com/doi/full/10.1080/17453674.2021.1941628
My name is Dr. Jeanne Franzone. I am an Orthopaedic Surgeon at Nemours Children’s Hospital in Wilmington, Delaware in the USA.

Dr. Richard Kruse (Orthopaedic Surgery), Dr. Michael Bober (Orthogenetics) and I are Co-Directors of our Multidisciplinary Osteogenesis Imperfecta Program. Although we are at a children’s hospital, we are able to follow our OI patients and families during the prenatal period, delivery process and neonatal care and up through the age of 35 years old.

Tell us about your experience!
On an annual basis, we are fortunate to see hundreds of unique OI patients. At our center, we perform upper and lower extremity reconstructive procedures and OI spine procedures. Over the past two years, one quarter of our patient volume has been in the age group of 18-25 years.

What is the difference between operating children and adults with OI?
There are principles of OI orthopaedic surgery that apply to both children and adults, but also certain differences. Regarding the differences, we consider the preoperative work up, surgical procedure and recovery. The preoperative workup for adults is often more complex and requires many medical subspecialties as adults tend to have more medical comorbidities and conditions that require coordinated multidisciplinary medical care. Regarding the surgical procedure, it is critically important to address the specific reason for which the surgery is being done. What exactly is causing the pain or functional limitation?

Adults tend to have more difficulty healing osteotomies than young children do, and it is important to tailor the surgery to each patient and his or her specific issues.

This impacts the postoperative recovery as well, as it is preferable to minimize the postoperative immobilization for adults. Adults often additionally have family and work responsibilities to consider. Another consideration for all surgeries is Pain management. By the adult years, many OI patients are familiar with what options have worked well in the past and it is helpful to communicate this information to the perioperative care team.

Do you sometimes refer to an adult specialist?
Yes – there are certain procedures such as a total knee replacement for which we may refer to an adult joint replacement surgeon. Our pediatric center is somewhat unique in offering total hip replacement procedures for young adults with skeletal dysplasia.

What are the pros and cons of having OI-specialists who can also operate adults?
We feel strongly that it is important to care for patients with OI as a whole person and to care for OI patients with a multidisciplinary team.

Should we advocate for having more pediatric surgeons doing surgery on adults with OI?
The topic of transition of care for young adults with OI is a very important one. Advocating for multidisciplinary medical and surgical homes for patients with OI into adulthood would help improve OI care.

Should it be a collaboration between a pediatric OI-surgeon and an adult specialised surgeon?
Collaboration of pediatric OI surgeons and an adult specialized surgeon can be very helpful.

Do you belong to any international networks for OI-surgeons? If yes - which?
Dr. Kruse and I have organized an International OI Surgeons Work Group, which is an international gathering of specialized OI surgeons representing 10 countries and 3 continents. The group conducts case-based discussions to discuss surgical care in a way that spans different healthcare systems and available resources and also conducts multicenter OI clinical research.
DR. MICHAEL TO,
University of Hong Kong and Shenzhen

My name is Dr. Michael To and I’m a paediatric orthopaedic surgeon, working both in Hong Kong and Shenzhen. I graduated from the University of Hong Kong in 1999 and became a paediatric orthopaedic specialist in 2006.

I worked in the Duchess of Kent Children’s Hospital and Queen Mary Hospital in Hong Kong. I started helping patients with OI during my specialist training in 2000 in Hong Kong. Since 2012, I extended my practice to Mainland China in a city called Shenzhen. I commuted almost every day between Hong Kong and Shenzhen to help develop a new hospital, the University of Hong Kong (HKU) Shenzhen Hospital, in Shenzhen. I started a multidisciplinary OI service in about 2014.

Tell us about your experience with OI!
I have managed over 1000 patients through various free consultations in remote areas and cities in China at which I did free consultations over the past 10 years. In my hospital, the University of Hong Kong Shenzhen hospital, I started to operate in 2014/15 and have treated roughly 500+ patients according to our hospital system. I have done roughly 620+ operations on OI at which majority involve single stage multiple-bone operations because the deformities usually are complex and involving both lower limbs. I also operated on OI scoliosis.

I also work closely with charitable foundations to seek fundings for the patients in need. Currently there are four foundations I collaborated with in both Hong Kong and Shenzhen.

In HKU, the skeletal dysplasia patients are mainly followed up by paediatric orthopaedic surgeons. Therefore, we have close relationship with both the patients and their family members. I operate on both children (age 0-18) and adults (age >18). The oldest OI patient I operated was >60. I cannot remember the exact age and she is now nearly 70.

What is the difference between operating children and adults with OI?
The difference is huge. Children’s fractures heal faster and surgical dissection is usually simpler. The operation time is usually shorter. But as they grow up, the dissection is more, blood loss is more and the operation time usually takes longer. The choice of implants are also different. In children, we use telescopic rods to accommodate the growth. In adults, we use locking nails instead. The scoliosis in children tend to be more flexible and may be easier to correct. But adults’ deformities are usually stiffer and more severe and the surgical dissection and recovery usually takes longer. But overall, the experience and outcome is usually very gratifying. 80% of my patients come from outside Shenzhen or Hong Kong. They travelled long distance to seek medical attention. Their problems may be fresh fractures or long standing deformities.

Do you sometimes refer to an adult specialist?
So far we have not referred or done any joint replacement surgeries for our OI patients. But I work closely with our adult trauma, spine surgeons and neurosurgeons as well. In my opinion, we cannot possibly know everything. We need to work with adult surgeons.

What are the pros and cons of having OI-specialists who can also operate adults?
In my opinion, OI is not just about bone fragility. It involves multiple organs and require the inputs from different specialties – dental surgery, ENT, geneticists, IVF, paediatricians, endocrinologists, rehab specialists.... Children are not small adults and certainly adults are very different from children. There should be a comprehensive care no matter if we are helping children or adults.

Many people may think treating OI is like treating adult osteoporosis, we as surgeons, we operate and let the endocrinologist or geriatricians look after the medical part. OI however, is a life-long disease. The relationships between the doctors and patients are lifelong. To me, everything started the moment the child was born. We as paediatric orthopaedic surgeons know about the
patients and their families very well so that we know what the best for the patients may be. As we become more subspecialized (knee, hip, hand, spine, or pelvic surgeons), we may miss out areas that we are not familiar with and we (superspecialists) may not be able to provide the most comprehensive care for our patients.

There are lots of medical advances every day, the skills and treatments in different subspecialties are also advancing. These skills and knowledge may be useful in the treatments of OI where there are still many unanswered questions. An OI specialist can help to connect dots!

**Should we advocate for pediatric surgeons doing surgery on adults with OI?**

I think there are advantages for paediatric orthopaedic surgeons to operate on adult OI patients especially if the patients have been followed up for a long period of time. But there are also issues related to the government policy, insurance, etc.

**Could one solution be a collaboration?**

Collaboration between adult and paediatric orthopaedic surgeon is a good model in helping patients with OI. The paediatric orthopaedic surgeons may have more experience looking after the patients for years and know them inside out. The adult surgeons have more experience in adults and in fact many of the skills in treating children and adults are very different. It is certainly a win-win for patients.

**Do you belong to any international networks?**

I am a member of the Scoliosis Research Society, Asia Pacific Orthopaedic Association, Hong Kong Orthopaedic Association, and SICOT. Currently, OI surgeons from North America, Europe, Asia including Richard Kruse, Pejin Zagorka, RJB Sakkers, TJ Cho and many others around the world have formed an International OI Surgeons Study Group and we meet regularly online to talk about OI patient management. Our last meeting talked about pelvic deformities in OI. This can definitely improve our knowledge.

**Anything you think we should have asked about?**

Over the past 20+ years of medical practice, OI is one of most challenging diseases I managed. Not just the long bone deformities, but the spinal deformities and many non-orthopaedic challenges. I think my patients have made me become a better doctor. I work closer with experts from different disciplines e.g. dental surgeons, prenatal diagnosis, paediatricians, IVF doctors... to provide better holistic care of the patients.

I also work with patient organisations, charitable foundations and governments to improve the patient care. Through these outreach services to remote areas, I have met many patients with rare bone diseases who may lack medical and financial support. I am also grateful to have the opportunity to meet and work with many patients and experts around the world.

Literature review: Fracture risk in adults with OI

Interview with Dr. Winnie Liu, Oregon Health and Science University (OHSU), Portland, Oregon, USA

I am an adult endocrinologist and I take care of patients at Oregon Health and Science University (OHSU) located in Portland, Oregon. My clinical and research interests are metabolic bone disease with a focus on OI after joining the BBDC (Brittle Bone Disease Consortium) as a co-principal investigator.

Who was behind the project and which methods were used?
We are a close-knit team at OHSU including Eric Orwoll, MD, Lindsey Nicol, MD, Melanie Abrahamson-Sommer and Riley Johnson. We also received input from other experts in the OI field. We conducted a thorough PubMed search of studies involving osteo-protective medications in adults with OI.

What was your research project about?
As an adult endocrinologist, I was motivated to understand fracture risks and the effectiveness of bone protective medications in adults with OI. Thus, in this review, we highlighted the elevated fracture risk in adult patients with OI and summarized current knowledge on pharmacologic treatment options for reducing fractures. Based on available data, we provided clinical frameworks for the timing of therapy initiation, medication options and assessment of therapeutic responses. Lastly, we noted significant gaps in knowledge and opportunities for future studies.

What are the existing therapeutic options for fracture risk reduction in adults with OI?
All the medications we currently use to reduce fracture risk in OI individuals were initially developed to treat osteoporosis. There are no FDA-approved bone strengthening medications for OI patients. Oral and intravenous bisphosphonates have been shown to improve BMD in adults with OI and are the most commonly used therapeutic agents; however, conclusive data confirming fracture protection are lacking. Teriparatide, a PTH-analog, appears to increase BMD, but thus far, robust increase in BMD seems to be limited to those with type I OI and fracture risk reduction remains unknown. We also touched on newer agents such as anti-sclerostin antibodies and TGF-beta inhibitors. More research is definitely needed, to further our understanding of these medications.

What is known about side effects (ONJ, atypical femur fractures etc)?
Osteonecrosis of the jaw (ONJ) and atypical fracture of the femur (AFF) are rare but serious complications of bisphosphonate therapy. The true incidence of bisphosphonate-related ONJ in OI adults is unknown, but based on available data, it is unlikely to be significantly higher than that of adults treated with bisphosphonate for osteoporosis. The link between bisphosphate treatment and AFF is difficult to established because AFF that develop in adult and pediatric patients with OI during bisphosphonate treatments may be due to inherent abnormality in bone quality due to OI rather than to bisphosphonate exposure.

What were your most interesting findings?
Our main conclusion is that bisphosphonate remains the mainstay of treatment but there are some promising data on PTH-analogs (teriparatide/abaloparatide). At the same time, it is important to note that bisphosphonates increase BMD, but we don't necessarily know that an increase in BMD translates into a decrease in fracture risk in adults with OI. We were surprised by the paucity of published data on the use of denosumab in adults with OI. Thus, further research is definitely needed. Additional trials are needed to examine the effectiveness of therapies in reducing fracture risk.
What is the most important take home message for clinical work?
The timing and duration of osteo-protective treatment should be individualized and we should pay special attention to older OI adults and postmenopausal OI patients. We should particularly consider treatment when there is a decline in BMD or new fractures. Bisphosphonates remain the mainstay of treatment, but teriparatide and abaloparatide can be considered for patients with type I OI who are intolerant or had inadequate response to bisphosphonate therapy.

How can we achieve more research in adults with OI?
Increasing knowledge of the gap in research involving adult OI is necessary. Close collaboration among clinicians and scientists who study OI are important to direct our research efforts and resources to the most pressing issues identified by individuals living with OI. I also encourage adults with OI to participate in research studies.

Were patients/patient organizations involved in this review? How?
We included a clinical vignette of an OI patient seen in our clinic to illustrate the importance of the questions that our patients have brought to us.

The whole article can be read here (open access): https://pubmed.ncbi.nlm.nih.gov/36658750/

Stem cells and Cell therapy (TERCELOI Project)

Interview with Clara Isabel Rodríguez, Biocruces Bizkaia Health Research Institute in Bilbao, Spain

My name is Clara I. Rodríguez and I’m the group leader of Stem cells and Cell therapy laboratory at Biocruces Bizkaia Health Research Institute in Bilbao, Spain.

Our laboratory focuses on the potential of human stem cells and their derivatives for the study of diseases and the development of new therapeutic strategies. The main research line of the lab is centered on the development of new efficient therapies for OI. We are developing several projects based on drug screening and repurposing in addition to the development of advanced therapy approaches. It is in relation to the latter that we have conducted a phase I cell therapy clinical trial (code: TERCELOI), demonstrating the safety and therapeutic potential of reiterative infusions of histocompatible BM-MSCs, in two OI pediatric patients. TERCELOI resulted in a decrease in the number of fractures, an improvement in bone parameters and the quality of life of the patients. The study of the mechanism of action indicated that MSCs therapy elicited a functional pro-osteogenic paracrine response in TERCELOI patients.

Tell us about the TERCELOI project!
The TERCELOI clinical trial as well as the cell mechanism of action studies was funded by the Spanish Ministry of Health through the call for independent clinical trials projects “EC10-219,” Instituto de Salud Carlos III through the project “PI15/00820” (Cofunded by European Regional Development Fund; “A way to make Europe”), local crowdfunding: (Bioef-EiTBI maratoia (BIO14/TP/007), and the AHUCE Foundation.
What was your research project about?
Mesenchymal stem cells (MSCs), as the progenitors of the osteoblasts, the main type I collagen secreting cell type in the bone, have been proposed and tested as an innovative therapy for OI with promising but transient outcomes.

The rationale for this approach was that after transplantation, the MSCs as the progenitors of osteoblasts, would engraft in host tissues, differentiate into functional osteoblasts, and secrete normal collagen type I. This would ameliorate the symptoms associated with OI, as has been previously suggested by the identification of mosaic individuals demonstrating that high proportions of mutant osteoblasts (carrying OI mutation) are compatible with normal skeletal function.

Notably, the first cell therapy clinical trials were carried out by Horwitz and collaborators, in which, after cell therapy (1 or 2 MSCs infusions) OI pediatric patients showed improvements in terms of growth velocity and fracture frequency, but these beneficial effects were transitory and the expected cell engraftment into bone tissue was anecdotal. It is worth to mention that these first clinical trials were performed in breastfed children affected by severe OI, who were previously subjected to immunosuppression. The detailed mechanisms underlying these beneficial effects of MSCs therapy in OI are still unclear, but given the low engraftment achieved in cell therapy clinical trials applied to OI, the possibility of a paracrine therapeutic effect of MSCs is gaining attention.

To overcome the short-term effect of MSCs-therapy and to avoid subjecting the patients to an immunosuppression process, we performed a phase I cell therapy clinical trial based on five infusions of histocompatible MSCs, in two pediatric patients affected by severe (patient P01) and moderate (patient P02) OI (code: TERCELOI). The aim of TERCELOI was to assess the safety and effectiveness of five MSCs infusions along 2 years in non-immunosuppressed OI pediatric patients. Besides, the host response to MSCs was studied by analyzing the sera from OI patients, collected before, during and after the cell therapy.

**TERCELOI outcomes:**

**Safety:** We first demonstrated that the sequential administration of MSCs was safe in both patients (five infusions each patient).

**Effectiveness:**
- Reduced the number the fractures (see annexes, figure A). Thus, in accordance with their severe (P01) and moderate (P02) OI, both patients had a significant number of reported fractures from birth and through childhood: 15 in the case of P01 and 11 in the case of P02. During the year prior to MSCs therapy, P01 had 8 more documented fractures, and P02 had 2. Remarkably, during the 2.5 years of cell treatment, P01 showed a reduction in the number of fractures (3 documented fractures), whereas P02 reported only 2 fractures. Moreover, this effect was maintained in both patients until the end of the follow-up visits (one and two years after the fifth and last cellular infusion).
- Increased mobility, improved the bone parameters, and quality of life of OI patients along the cell treatment plus 2 years follow-up period.
Moreover, the study of the mechanism of action, indicated that MSCs therapy elicited a functional pro-osteogenic paracrine response in the TERCELOI patients, especially noticeable in the patient affected by severe OI (patient P01), demonstrating its functional capabilities. A major finding of our study, from a mechanistic point of view, is the existence of a systemic functional pro-osteogenic response in OI patients as a consequence of MSCs therapy.

What is the most important take home message for clinical work?
For a laboratory researcher like me, it has been incredible to work so closely with the clinicians involved in the daily care of OI patients. I choose as the most important lesson I learned from this experience is that clinical and research work should move hand in hand to figure out the best way to improve the quality of life of the OI patients and their families.

Were patients/patient organizations involved?
It had been inconceivable to carry out the clinical trial TERCELOI without the involvement, support and trust of the patients and their families. The Spanish OI organization AHUCE was crucial to contact and recruit the candidate families along the country, in addition to financially contributing to this work. Five families met the initial criteria to perform the histocompatibility analysis (that consists of checking the immunologic similarity in blood samples) between the patient (children >6 months and <12 years old) and their healthy siblings. Eventually only two patients could be included in the clinical trial. The participation of the healthy siblings with the bone marrow donation was absolutely courageous, and the engagement and joyfulness of the young patients was inspiring during the whole process. I would like to express my deepest gratitude.

Stem cell therapy is regarded as an advanced therapy medicinal products (ATMP) - are there any special challenges regarding access to these therapies for patients?
TERCELOI patients’ treatment required the donation of bone marrow-MSCs from an histocompatible sibling, which implied the need to have a healthy histocompatible sibling to donate stem cells. Not all pediatric OI patients who were able to benefit from this therapy possess a histocompatible donor, which definitely restricts their access to this therapy. In this regard and given the encouraging clinical and molecular outcomes of the TERCELOI clinical trial, we hope to increase the effectiveness of the cell therapy and to make it available to a larger number of patients without the need for a histocompatible donor. Our laboratory is working on a new line of research focused on the therapeutic potential of MSCs derivatives, such as the extracellular vesicles (EVs), which in addition of avoiding the need for a histocompatible donor, provide an abundant source of medicine, able to being repeatedly administered, and can be enriched to increase their effectiveness.

You can read the articles about the project here:  
Who are you & what is your relationship to OI?
My name is Gisele Martins and I’m a professor of nursing at the University of Brasilia (UnB), Brazil. My involvement with the OI community started seeing OI kids with bladder and bowel issues in my advanced nurse-led clinic in the field of Pediatric Urology, located in our teaching hospital. Given my nurse-scientist background, I decided to conduct a preliminary study to estimate the prevalence and presentation of bladder, bowel, and combined bladder and bowel symptoms experienced by Brazilian children with OI. This research was published as follows: Martins G, Siedlikowski M, Coelho AK, Rauch F, Tsimicalis A. Bladder and bowel symptoms experienced by children with osteogenesis imperfecta. J Pediatr (Rio J). 2020; 96:472-8.

Who was behind the project and which methods were used? How was it financed?
This first preliminary Brazilian study was led by my research team at the University of Brasilia, Department of Nursing along with a collaboration between McGill University and Shriners Hospital for Children, Montreal, Canada. At that time (2015), Dr. Argerie Tsimicalis and I conducted a descriptive study with a convenience sample of 31 Brazilian parent-child pairs of toilet-trained children aged from 3 to 18 years. There was no funding.

In 2018-2019, I spent a bit of my sabbatical year under Dr. Tsimicalis’ supervision to conduct a mixed-method study which allowed for a comprehensive and rich account of the state of bladder and bowel issues in children living with chronic musculoskeletal conditions such as arthrogryposis multiplex congenita (AMC), Osteogenesis imperfecta (OI), and spastic Cerebral Palsy (CP). The study was grounded in the perspective of key stakeholders, including children, their family, and healthcare providers. It was funded by Shriners Hospital for Children and the findings were not published yet.

What was your research project about?
It was about bladder and bowel issues in children living with impaired physical mobility. The first preliminary study was conducted with a Brazilian sample of 31 children with OI and the second was composed by 45 Canadian children living with impaired physical mobility, including children with OI. A detailed understanding of the continence issues, the challenges associated with toileting practices and barriers related to public access will help individuals, families, and healthcare providers to advocate for treatment, services, devices and environmental modifications.

What was your most interesting findings?
Speaking of children with OI, constipation was the prevalent symptom in both samples (Brazilian & Canadian). Moreover, a combination of bladder issues (such as holding maneuvers and urgency) and bowel symptoms (such as hard or painful bowel movements and large diameter stools) was also reported, while isolated bladder issues were not reported by our samples.
What is the most important take home message for clinical work?
This research is an important step toward effective screening, detection, and access to care and treatment, especially for individuals living with impaired physical mobility.

Do you think the findings are relevant for adults with OI as well?
The OI Foundation has identified underemphasized health conditions in adults with OI “urinary tract” or “gastrointestinal system” as the organ systems that had the most significant negative impact on their quality of life. Additionally, early identification of gastrointestinal problems and a timely referral to a specialist is crucial to avoid these symptoms throughout the life course.

Argerie (Tsimikalis), can you tell us about the pooping video?
Gisele’s work in Brazil, followed by our work at Shriners together (unpublished) found that some of the children are constipated. Now, the risk of constipation is greater after surgery. Being constipated is not very pleasant, not very pleasant to talk about and can even have some embarrassing moments.

Now, how does one talk about poop? As part of our effort to increase to educational resources available, our team has created a colouring book for children with OI called OI Colour OI Learn. The book is supplemented with colouring sheets, greeting cards and animated videos. Learning how to have a successful poop after surgery, is one of the animated videos. Why animation? Makes it fun, adds humor, help destigmatize, and welcomes children to talk about poop and farts, and learn what is needed to make those Bowls move. Link to video: https://youtu.be/yw0eajzMC0w
Should more research be done on this topic?
Conservative interventions, including lifestyle changes and behavioral therapies, have not been examined specifically in children with OI experiencing bladder and bowel issues, and represent an optimal opportunity to provide relief and improve quality of life.

Were patients/patient organizations involved in this project? How?
Absolutely! Our team is composed of scientists, healthcare providers, and patients to facilitate and mobilize research evidence in the hopes of increasing awareness of bladder and bowel issues, particularly in children living with impaired physical mobility.

Any messages for the readers of OIFE Magazine?
Advocate for bladder and bowel assessment across lifespan. It is also important that healthcare providers, parents, and when appropriate the child, can identify the red flags of incontinence, such as delayed toileting training, urgency associated with ‘accidents’, soiling, and continually monitor bladder and bowel control with a low threshold for referral.

Artist with OI: Athena Cooper

Athena Cooper is a visual artist and creativity coach based in Calgary, Canada. Self-taught as an acrylic painter, Athena's paintings are explorations of what it means to live an extraordinary, ordinary disabled life.

Please tell us a bit about yourself!
I'd describe myself as someone who has had a pretty wide-ranging set of experiences and adventures over the course of my life thus far. I was born in Vancouver, Canada and I trained as an animator once upon a time, but then spent about the first 15 years of my career working in web development and digital marketing. I left Vancouver for the prairie city of Calgary, (just east of the Rockies), back in 2018 and then left the corporate world a few years later, to strike out on my own as a visual artist and creativity coach. My husband and I love the life that we're continuing to build in Calgary. We have an apartment downtown not far from the Bow River and take frequent walks through the neighbourhood’s green spaces with our two small dogs, Lucy and Lola. He and I also have a family practice together called the Tilted Windmills Healing Centre where we use creativity as a tool to help improve mental health and well-being.

I have moderate to severe OI, (likely Type 3 or 4 but I've never been tested). This means that I do require a power wheelchair for mobility and safety, however, I am able to transfer independently and do things like stand on the footrests to do dishes or climb in and out of the bathtub.

I would say that the biggest impact on my daily life is just getting around. Calgary's winter begins in October and there can be snow on the ground all the way into April. It's prairie snow so it tends to be quite light and powdery, but it does build up over time. One of the reasons we live downtown is that the sidewalks get cleared of snow fairly quickly. I also use a door-to-door disability transit service to take me to and from my studio space during the week.
Can you describe how you are working? Are there particular challenges due to your disability?

I've been painting with acrylics on and off since high school, however, I got more serious about my painting practice a little over ten years ago. I was struggling with my creativity at the time, and really wanted to get away from working on the computer which dominated my day job. I noticed that I was collecting a lot of images of Tiffany stained glass on Pinterest. I decided that I wanted to try doing paintings of stained glass lamps and panels in acrylics to learn more about why I found them so fascinating and beautiful. I've since moved on to painting a range of landscapes, still life and portraiture, but that sense of intense colour and fractured light of stained glass is still very much a signature of my artistic style.

OI impacts my art in the fact that I can't paint for very long before I start to fatigue or the scoliosis in my lower back starts to bother me. I typically paint for about an hour and a half to two hours a day, which means that a single painting will take anywhere between a week to two weeks to complete.

I also paint on quite small canvases. I find I can't manage anything much larger than 11x14" or 12x12" as it requires too much reach and I'll fatigue even more quickly. For my current project, I'm working on nothing but 8x8" canvases.

Tell us about your 2024 art exhibit and the "Love & Disability" online Survey!

While OI has offered its health challenges, I felt these tended to dominate other people's perceptions of my disability more than my own. They have these assumptions about my level of pain or my level of physical hardship that doesn't feel true to me. On the other end of the spectrum, there's this popular narrative about being inspirational, simply for living a life that includes disability that also doesn't sit right.

Through my art, I often explore the theme of what it means to live an ordinary disabled life. I don't see my life as exceptional because of my disability, and this shows up in my paintings through my depictions of the everyday such as places in my neighbourhood or a string of espresso cups in a sunbeam. One of my favourite paintings, "Trusty Steed", shows my view of my power wheelchair as I sit on the grass at a music festival. The wheelchair however has no more positive or negative emotional charge than a piece of furniture.

For my art exhibit, "The Extraordinary, Ordinary Nature of Interabled Love", I will be highlighting the everyday moments in my relationship with my husband from our first meeting nine years ago all the way through the pandemic and new puppy adventures. I really struggled in my early adulthood to believe that I would ever find a romantic partner. Part of my insecurity stemmed from not seeing people who looked like me in relationships. The exhibit is in many ways the representation of an ordinary disabled love story that I wanted to see when I was younger.
I also feel it's important to extend this narrative beyond that of myself and my husband. This is why I am inviting other couples where one or both partners identify as disabled to also contribute to the exhibit through the “Love & Disability” online survey. The survey is primarily made up of four open-ended questions in which participants are invited to talk about their relationship. Submitted photos and portions of their responses may be selected to be incorporated into the exhibit itself, which will be happening in Calgary in the late summer of 2024.

I know everyone’s love story is different and I really want to share all the ways that couples have found ways to support one another and thrive.

**RELATIONSHIPS AND OI**

When Athena Cooper from Canada was younger, the lack of representation of people with disabilities in romantic relationships filled her own search for a partner with doubt and insecurity. Nine years after first meeting her husband, she is now exploring this subject through an exhibit of her paintings set to open in the summer of 2024.

As part of this project, she is inviting couples to share their own experiences of love and disability through an online survey. There’s also an opportunity (entirely optional) for people to contribute their photos as well.

If you’d like to support, please visit the online survey at [http://bit.ly/LoveAndDisability](http://bit.ly/LoveAndDisability) to share your experience or pass this link on to anyone who you think might be interested in participating. She will be collecting submissions until May 31, 2023.

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**Welcome to the newly founded ISCBH!**

The ICCBH are now the International SOCIETY of Children’s Bone Health, and they are ready to accept members!

The International Society for Children’s Bone Health is a scientific society dedicated to achieving the best bone health for every child across the globe through research, education, collaboration and advocacy. Emerging from the long-running ICCBH we are now a fully-fledged society in our own right and our Society has been created to meet the emerging needs of the children’s bone health community.

You can find full information and submit your membership application on their new [website](http://www.iscbh.org). Membership lasts for 2 years, as the cycle of our main conference is biennial.

Benefits of membership include access to educational resources, great discounts on participating in their meetings and conferences, eligibility for grants and awards, updates and news from the field but most of all you will be part of their wonderful community!

You can join online [here](http://www.iscbh.org), and if you have any questions or need to get in touch you can do so on their new email address: [info@theiscbh.org](mailto:info@theiscbh.org).
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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<tr>
<th>Statements about severe types of OI (type III and severe type IV*)</th>
<th>Yes</th>
<th>No</th>
<th>Comment, if any:</th>
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<td>During the first 2 years of age the fracture incidence decreases with age</td>
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<td>During the first 2 years of age the fracture incidence is stable over time</td>
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<td>2 Will fractures increase after weight bearing / moving actively on their own (crawling, bottom shuffling, walking etc)?</td>
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<td>3 Select one time point</td>
<td>&lt;9 months</td>
<td>6–18 months</td>
<td>18–30 months</td>
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<td>Approximate age when you start rodding surgery?</td>
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<tr>
<td>4 Select one choice</td>
<td>&lt;5 years</td>
<td>5–10 years</td>
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<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
CLINTEC | Karolinska Institutet
+46 070-471 23 00 | +46 8 524-833 95
KI.SE | BOOSTB4.EU
https://staff.ki.se/people/cecgot
EUROPEAN HEALTH DATA SPACE (EHDS)
OIFE completely agrees with EURORDIS and the other signatories that the interests of people with rare conditions and other health problems needs to be taken into consideration when developing the European Health Data Space. We cannot make sharing of data too complicated or leave it into the hands of commercial stakeholders. To advance knowledge on rare conditions - we need to share data between European countries. We fully support the joint statement to the EU Policymakers. Read the joint statement here.

THE CURRENT STATE OF OI RESEARCH
OIF SCIENTIFIC MEETING RECAP

NEWS FROM OI RESEARCH
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BEER FOR OI
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4FRAGILITY PROJECT - GAIT ANALYSIS AND HR-pQCT IN ITALY
The IRCCS Istituto Ortopedico Rizzoli has started recruitment of patients to two clinical studies. The Italian organization ASITOI is helping with recruitment. The first 4Frailty project "Intelligent sensors, infrastructures and management models for the safety of fragile patients" - involves the use of an advanced movement analysis system ('gait analysis') in people with OI. Gait analysis is used to measure the kinetic and kinematic characteristics of body movement during walking or other simple exercises of daily life (such as getting up from a chair, climbing a step, etc.).

The second project is open to all patients affected by rare skeletal conditions and includes bone evaluation using High Resolution peripheral Quantitative Computed Tomography (HRpQCT) - a tool to evaluate both qualitative and quantitative variations of the bone. The objective of this project is to compare dual energy X-ray bone densitometry (DXA) with HRpQCT.

ORTHOPAEDIC TREATMENTS OF OI ADULTS
The OIF has published a video of a webinar about orthopaedic treatments of adults with OI hosted by the Orthopaedic Trauma Association: https://youtu.be/NiccYsS_ElI

The session helped attendees:
- Understand the current gap of treatment in caring for adults with moderate/severe OI
- Learn basic surgical treatment principles in managing patients of all ages with OI
- Identify typical pediatric implants in moderate to severe OI patients to facilitate transition of care in adulthood
- Learn basic concepts of surgical problems and surgical care in adult patients with OI.

BRONZE STATUE OF STELLA YOUNG
Two months before she died, Stella Young – comedian, writer, disability activist and self-described “crip” – wrote on Facebook how she wanted to be remembered. “I am not a snowflake. I am not a sweet, infantilising symbol of fragility and life,” she said. “I am a strong, fierce, flawed adult woman. I plan to remain that way, in life and in death.” March 30th a bronze statue of Young in her wheelchair by sculptor Danny Fraser was unveiled in the disability activist’s home town of Stawell, Australia as part of the Remembering Stella Young project. Read more here.
FUNDING FOR OI-RESEARCH
Are you a researcher or do you know an OI-researcher with a good idea, but no money? Tell them to check out the grant from the Brittle Bone Society (UK & Ireland)! Researchers from outside UK can also apply (deadline May 31!)

VR AGAINST PAIN AND ANXIETY
At Shriners Hospitals for Children Canada they have started to use VR in an effort to distract, before medical procedures that can cause anxiety in children with OI. They put VR headsets equipped with a video game on children undergoing medical procedures that could cause anxiety and pain. The researchers say the results were almost exclusively positive. Kids get completely distracted by the immersive game. Dr. Reggie Hamdy appreciates how unlike anxiety-reducing drugs, there are no side effects from playing a VR game. Read more here.

What is the GloBE-Reg?
GloBE-Reg is an international registry project. The stakeholders of this project include patients, professionals, scientific organisations and industry.

Industry-led registries for post-regulatory approval studies of safety and effectiveness have limited long-term sustainability which compromises their utility for collecting long-term outcomes. The GloBE-Reg project was launched in early 2022 with the aim of developing a registry platform for post-marketing surveillance studies. The platform has a common core data set which complies with international guidance for rare disease datasets and has sufficient versatility to allow inclusion of any drug class and brand for development of a diagnosis and drug specific clinician reported and patient reported outcome module. The platform and the registry received ethics approval in the UK in late 2022 and is now extending to other centres.

One of the first tasks of the GloBE-Reg project was to develop a drug-specific module to collect information on rhGH (recombinant human growth hormone) therapy in children. With the introduction of new indications and novel forms of rhGH, the need for a global registry had never been greater. This work was supported by a GH Scientific Study Group which developed a minimum dataset for collection in all children receiving rhGH therapy for GH deficiency (GHD). This process can now be used to collect information on other types of rhGH therapies for any indication.

The project management team of GloBE-Reg is based at the Office for Rare Conditions at the University of Glasgow and the project, itself, is solely dependent on industry support. With its initial focus on rhGH therapy, its current funders are GenSci, Novo Nordisk and Pfizer. The project is guided by a Steering Committee and a Data Access Committee and a GloBE-Reg Patient Advisory Group. The process for developing the GH Study Module has shown that the GloBE-Reg structure can support the needs of a wide range of patients, health care providers and industry. To find out more, please visit the Registry (globe-reg.net)
Announcement Pega Medical

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OIFE Topical Meeting in Stockholm – streaming tickets now available!

OIFE and the OIFE MAB invite you to register for the 2nd virtual European Investigator Meeting for osteogenesis imperfecta (OI) on November 17th from 14 – 19 CET.

Do you want to be informed about the event? Please sign up here.

Contact & Calendar

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

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

Website: http://www.oife.org
Facebook: https://www.facebook.com/oioife
Twitter: @OIFE_OI
Instagram: https://www.instagram.com/oioife/
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