Editorial

By Ingunn Westerheim, OIFE president

Tired, fatigued or just lazy...?

It’s not something I do very often. But every time I’m moving to another apartment, I’m thinking “never again”. It’s close to a returning nightmare. Needing help with most of the practical stuff because of my disability, doesn’t make it any better. For every box that needs to be moved or carried, every lamp that needs to be installed, every curtain that needs to be hung and every piece of furniture that needs to be unwrapped and put together, I need to ask someone else for help.

It’s stressful in itself. Fortunately we had a lot of good helpers this time. We had two independent living assistants paid by the town and a moving agency to help with the heavy stuff. And I got tired just from watching...
This time we moved to a newly constructed apartment, where nothing was in place. In addition came all the paperwork. And at some point, I found myself falling into a big black hole. One week after moving in, I was falling asleep at the dinner table and the only thing I could think about was my bed. Every day that went by I was more tired. It was like a spiral or a dark hole I was falling a little bit deeper into for every hour and very day.

I spent more and more hours in bed, but sleeping didn’t help! After going to bed before 8PM, I woke up 4-5 hours later. Because of lacking curtains, a new mattress, pain, stress and the thought of everything that needed to be fixed the next day.

After two weeks in this state I felt more or less like a zombie. I had aches and pains all over and poor breathing. So I called my doctor and told him that my OI-body was about to collapse, and that I needed some days off. My OI-body was not working with me anymore. It was working against me.

Fortunately, my family doc has known me for 25 years. He knows that I don’t skip work easily. He agreed that I should take it easy for the next 2 weeks. Afterwards I went to see my massage therapist and he found a lot of trigger points. Pain was radiating into my arms and fingers. That night I took a muscle relaxer and went to bed early. And I slept for 13 hours. Deep good quality sleep. Finally!

The next day I was still not recovered. But at least I felt more human than zombie. So what was the problem? Sleep quality? Pain disguised as tiredness? Stress? Laziness? Or fatigue? This complex word I have a very ambivalent relationship to. Because what does it really mean? And in what ways is it different from tiredness? And do people with OI struggle more with it than other people?

According to the study of dr. Heidi Arponen, included in this magazine, the answer to that is no. But could it be because the way the questions were asked? I have a strong feeling that people with OI struggle more with these issues than other people. But nobody has managed to document it in a convincing way.

Because every human on earth feels tired sometimes. It’s completely normal. But do they really feel fatigued? Who knows? Maybe life is stressful for everyone? Especially now with Corona, where many factors providing quality in life are put on hold and SoMe and news are driving us crazy. Is everyone fatigued to some extent?

Honestly, I feel there is a still a piece in this puzzle missing. I think people with OI are tired in a different way than other people. Both mentally, but first and foremost physically. And it’s probably a combo of overperforming at work, education and daily life activities, because “we want so hard to be normal”. Our muscles and lungs are poorer quality, which causes less endurance and poorer breathing. We also have a lot of suppressed pain we’re not even aware of.

Pain can create poorer sleep quality and make us less rested. The Finnish study also showed that a number of people with OI have sleep apnea without knowing.

We clearly need more understanding of the mechanisms behind the different kinds of OI-pain and if they cause fatigue or not. These are one of the important messages we as patient representatives bring into the construction of the OI-module in the new European OI-registry (EuRR-Bone). In addition to research and registries, we as patients must be willing to provide the necessary data through surveys, signing up for registries and using wearables and other new and innovative tools.
But people are not the only ones who get fatigued. The last months I have realized that a number of the OI-organizations are struggling. It’s hard to engage people when face to face meetings is not an option. It’s hard to raise funds, host AGMs and to recruit and elect new boards members. Too many organizations are dependent on that one engaged person. When that person has lost the spark or suffered a burnout, the group might be left in a very difficult situation.

The OIFE is aware of this and we take it seriously. We are reaching out to our members asking if there is anything we can do to help. My advice is to work systematically with recruitment of new members, volunteers and board members throughout the year.

Find out who your members are and what they are good at/interested in!

We should also be better at providing positive feedback to those people who bother to do the work, who keep the wheels turning and who takes one for the team. Sometimes leading an organization can be a very lonely job. Make sure to give a pat on the shoulder to people who take on that job! And ask if you can do something to help. Sometimes it only takes a smile or a positive comment to motivate someone to keep on keeping on.

Hang in there and stay safe everyone!

Ingunn – OIFE president

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

First of all I want to start by congratulating the OI Foundation with their 50th anniversary. On August 21st it was 50 years since our partner organization in the USA - (OIF) - was founded. August 21st we accidentally discovered this treasure when going through boxes in storage. The plaque was a gift from the OIF to the BBS at one of the first international meeting for OI-organizations in Scotland in July 1982. This was many years before OIFE was established, 27 years ago. But collaboration across borders started early, and is more important than ever. We’re sorry that your big celebration has to wait because of Covid. But we’re looking forward to celebrating with you when international travel is possible again. Until then we will enjoy the recordings from your virtual conference from July 10th – 11th:

www.oif.org/virtualconference!
Meetings and events

Videocalls in the last months have included meetings related to ERN BOND, Mereo Biopharma and ICCBH Virtual Forum, where OIFE and OIF are formal collaborators. Meetings also included videocalls in The CHRONOS project, where OIFE is part of the consortium. There have also been videocalls (VCs) with OIFE members, volunteers and 4 VCs with individual members of OIFE MAB in addition to:

- EC-meetings on Zoom July 21st, Sep 22nd and Oct 20th
- Several webinars connected to EURORDIS Digital School (AR)
- VC with European Joint Programme on Rare Diseases June 22nd (IW)
- Webinar on Innovative Medicines Initiative (IMI) Call (IW)
- VC Pega Medical July 8th (IW, DL and BvD)
- VC Austrian researchers about Photovoice project August 4th (IW)
- VC with working group of US OI-surgeons on case discussion project Aug 5th (IW)
- VC with company Longenesis Sep 7th (IW and RTS)
- Webinar Key4OI update Sep 9th (IW)
- ERBF Webinar Genetics of Rare Bone Diseases Sep 17th (IW)
- VC - establishment of Mereo survey steering committee Sep 21st (IW and TvW)
- VC with dr. Joan Marini Oct 1st (IW and Tracy Hart)
- VC between OIFE EC and the Board of Care4BB Oct 7th (all EC-members)
- Webinar on Strategies for Annual Fundraising Oct 27th (BvD)
- ERBF webinar QoL-research Oct 29th (4 patient representatives represented OIFE)

A stronger BOND between us

XLH Alliance & OIFE

X-linked hypophosphatemia (XLH) is a genetic condition characterized by low levels of phosphate in the blood. Phosphate is abnormally processed in the kidneys, which causes a loss of phosphate in the urine (phosphate wasting) and leads to soft, weak bones (rickets).

This means XLH has a completely different cause than OI. But in spite of this, people with OI and XLH struggle with many of the same challenges. Fractures and surgeries as a child. And pain, fatigue and mobility issues as an adult. We also share problems with teeth and hearing. With the kind support from Kyowa Kirin International (KKI) OIFE has developed a formal collaboration with the XLH Alliance to learn from each other and to support the XLH Alliance, who’s in a phase of enlargement.

Ingunn has hosted two meetings with Oliver Gardiner and Tenna Toft from the XLH Alliance on July 5th and Sep 7th (IW). And from November we are planning monthly meetings to share ideas, best practises and potential projects we could work together on.
OIFE at BBS Conference
OIFE also had several people attend the Annual Conference of the Brittle Bone Society. The conference included talks about mild OI, women’s health, diet & nutrition, updates on OI-research and more. You can find recordings from all of the sessions through this link: https://bit.ly/3dGkN7l

Patient Engagement Open Forum
Representatives from OIFE have attended several of the session of the Patient Engagement Open forum – a series of webinars from June 25th to November 2020. The topics have ranged from tools and recommendations for effective patient engagement, methods for monitoring and evaluation of impact and outcomes in patient engagement activities, and fair market compensation for patient input to interactive sessions on assessing good practices in patient engagement and more. One of the webinars was on drug pricing and what mechanisms lies behind it. Across Europe there is disparity in the average time for new drugs to be approved and accessible to patients. There are a number of factors delaying these processes. Typically, a pharmaceutical company will initially seek centralised approval from the European Medicines Agency (EMA) for a marketing authorisation. This approval, if granted, demonstrates that the EMA believes that the medicine is safe and effective and can be sold within Europe. However, it does not agree on, or decide the price of the medicine. Manufacturers must still negotiate over price with each country on an individual basis. Country approvals can take a long time and there are big differences from country to country. For an orphan drug, the delay between EMA approval and in-country commercial availability varies from 39 days in Germany to 1,236 days in Poland!

EuRR-Bone & Summer School on Registries
Ingunn Westerheim and Rebecca Tvedt Skarberg have represented OIFE in the OI working group of EuRR-Bone which had their first meeting October 29th. To be better prepared for the task we had two meetings with Natasha Appelman-Dijkstra beforehand. Dagmar Mekking also attended the working group meeting on behalf of Care4BB & Key4OI. The so-called Vertical Track working group is led by dr. Wolfgang Höegler from Austria and will make a suggestion for PROMS and CROMs in the OI-specific module of EuRR-Bone. Read more about EuRR-Bone here: https://eurr-bone.com/

Ingunn also attended the Summer School on Registries, which took place virtually from October 28th – 30th. Coreen Kelday attended the last sessions of the training on behalf of Ingunn. Registries and patient reported data was also the main topic on the Norwegian organization’s autumn meeting, where Ingunn represented OIFE.

OIFE supports EU4HEALTH campaign
OIFE has joined EURORDIS and signed the campaign "Europe, Let’s Do More for Health", which calls for targeted and effective EU action to ensure that everyone is able to enjoy healthy lives in healthy environments. The organizations call for an EU leadership that respects EU treaty provisions on health, as well as its international commitments. The EU and its Member States should cooperate to effectively address the unprecedented health challenges they are facing.
Meet the OIFE delegates

Coreen Kelday, United Kingdom
I work as the Support Development Officer for the Brittle Bone Society (BBS). We support people with OI throughout the UK and Ireland. I have worked with the BBS for 9 years and have been the OIFE delegate for 7 of those years. Prior to joining the BBS I worked in Private Healthcare for 10 years. I have a degree in Business Administration. During my spare time, if I am not spending it with my children and grandchildren, I am a keen musician playing both Flute and Piano.

The BBS was founded in 1968 in the town of Dundee in Scotland which is where we still have our HQ. We estimate there are around 5000 people with OI living in the UK and Ireland, and we have a database with around 4000 contacts. Our Charity receives no Government funding but rely on tremendous support from various Trusts, grant giving organisations, fundraising at corporate level and from our wider general membership.

We have 4 full-time members of staff; our Chief Executive is Patricia Osborne who is responsible for the day to day management and strategic direction of the charity. The Charity is governed by a Board of Trustees which consists of individuals from a variety of backgrounds with expertise in: Legal, Accountancy, Human Resources and Healthcare professionals. Many of the trustees have OI themselves or a member of their family does.

The BBS has a Medical Advisory Board established in 2012, who meet regularly alongside our CEO. We also formed a Scientific Advisory Board (SAB). In 2016 we gained membership of the AMRC (Association of Medical Research Charities). Our Charity has now funded 5 pilot research projects into OI.

As Support Development Officer I also work closely with POINT (Paediatric OI National Team), which is a working group of specialist paediatric allied healthcare professionals with an interest in OI. Both the MAB and POINT offer invaluable support with a wide range of activities from: providing healthcare updates and providing speakers for our events, writing and updating our factsheets, they also provide a steer with support enquiries.

Since 2015 we have been actively calling for establishment of a national (multidisciplinary) health service for adults with OI. In doing so, we have developed good relations with other rare bone groups such as HPP, XLH and Fibrous Dysplasia. The BBS offers a variety of events for all age groups, handling Support Enquiries and educational resources, a large part of our work is raising funds for specialised wheelchairs and equipment. On average we fund £100,000 per year towards Wheelchairs.

National Organisations have many important decisions to undertake, the most important is to support their own members at grass root level and to be advocates for their community.

If I was president of OIFE for a week I would probably look at opportunities for securing funding to pay a salaried member of staff. I congratulate the volunteers who have kept OIFE running for over 25 years without the input and support of employed staff. Many of the volunteers are also holding down full-time jobs and trying to run their own National Charities. It’s a tall order!

As an umbrella organisation for European OI patient groups one of the more important tasks for OIFE should be to coordinate what is happening within Europe on a wider policy level. In particular, in the current climate with increased interest from Biopharma’s and research into rare bone this is becoming increasingly vital.
Andrea Kiel, Germany

I’m 54 and living with my family (husband and two children) in Bavaria, near Munich. My two children are Aisha (22 years) and Yannick (19 years). Yannick has OI Type 3.

Shortly before Yannick’s birth, we were already on the way to Brussels to live and work in Belgium. My husband got a good job offer, and I was again on maternity leave. One year earlier we came back from Geneva and thought when our second baby is born we will repeat living abroad for a while. But life is not always as straightforward as you think.

And with Yannick’s diagnosis, which was not promising at all, we decided to stay in Germany. Plans changed and we had to adapt. We got a lot of help from doctors and our family and after a while we found a good way for our daily routine. For sure our life would have been looked different without OI. But would it be better? The biggest hurdle has to be taken by Yannick. We just try to help him find his space in the society and living a satisfying life without too much pain.

In OIFE, I represent Deutsche Osteogenesis Imperfecta Gesellschaft (DOIG), Germany, seated in Hamburg. The DOIG has more than 1000 members with OI and their families and is managed by volunteers. Since July 2020 the DOIG has a Director. Her name is Andrea König-Plasberg.

Ongoing projects/activities:

- The DOIG is offering an online Yoga course for all members. Together with a Yoga teacher we are working on a program which also should beneficial for people sitting in a wheelchair and without too much strength in arms and legs. For that reason, we also got in touch with the President of the Indian organization (IOIF).
- Working together with the ERN-Bond on an Emergency Action Plan for handling people with OI when infected with Covid-19.
- Annual weekends for members on a national and regional basis, acting as meeting point and for talks & workshops on OI-topics.

When I’m not doing OI-work, I’m a Food Technologist. I work in a Regulatory Affairs department of a smaller company producing fruit preparations and smoothies. Before I was working for an international company in a European R&D Centre in Munich, developing new products as chocolate or chewing gum and improving manufacturing processes.

My hobbies are reading books, swimming in the lake (during summer) and I love to travel, learning more about different cultures and admire the beautiful landscapes of our earth.

In my opinion the most important task for the national organizations is to make sure people have an address to connect, to exchange experience, getting tips and help in all kind of circumstances. To give the feeling of not being alone. To represent the needs of people with OI also outside the organization. Finding new friends.

OIFE should focus on the exchange of information and experiences on international basis. Networking. Learning from each other. Seeing things from a different perspective. Collecting and communicate information in a broader way.
Who are you & what is your relationship to OI?
I am a physician and rheumatologist with a special interest in the treatment of bone diseases. I have been taking care of patients with OI for many years and run a specialist clinic for these patients in Edinburgh at the Western General Hospital where I work.

Can you tell us how the TOPaZ project was initiated?
The project was initiated partly through my own desire to generate a better evidence base for the treatment of OI and also through conversations with OI patients who were surprised to hear that we weren’t sure whether any of the drugs that we use to treat OI are effective at preventing fractures!

This led to discussions with other stakeholders interested in OI. This included Prof Bente Langdahl in DK who also has an interest in OI, and the Brittle Bone Society in the UK. We were all concerned to try and improve outcome and that lead to me writing a grant to evaluate the effects of treatment on bone fractures in adults with OI. The study has been financed by the Efficacy and Mechanisms Evaluation (EME) board in the UK which is supported by the MRC and NIHR. We also received a donation of Teriparatide from Eli Lilly and company who provided the medicine free of charge.

What is the aim of the study?
The aim of the study is to determine if a 2-year course of teriparatide (TPTD) followed by an infusion of zoledronic acid (ZA) is superior to standard care in preventing fractures in adults with OI. We chose to study the effects of treatment on fractures since this is one of the most important complications of OI. It may be surprising to your readers that until TOPAZ, no trial has been designed to assess the effects on fractures. Rather, they have only studied effects on bone density.

To do a fracture prevention trial, quite large numbers of patients are needed. We have calculated that we will need to study 380 people with OI for 5 years to determine if the TPTD/ZA has a significant advantage over standard care. By that I mean that we are hoping TPTD/ZA may reduce fracture numbers by 25% or more than SC. At the moment we have enrolled 247/380 people so we are at 65% of our target. Standard care may consist of calcium and vitamin D supplements and bisphosphonates or denosumab or no treatment. Anything except TPTD. The study commenced in 2018 and has been in progress 2 years. We were hoping to end in 2023 but things have delayed a lot with Covid19 and I suspect it may be 2024 until we get the results. Having said that we are restarting recruitment now in the UK again and hopefully other countries will follow. Countries involved are UK, Republic of Ireland, Netherlands (Amsterdam), France (Paris) and Denmark (Aarhus), who are all in the planning/starting phase of recruitment.
The methods are to invite patients to the study, check a DEXA, spine x-ray (to assess vertebral fractures) and some routine bloods and questionnaires to assess things like pain and quality of life. Patients are also offered genetic testing to define the gene mutation responsible for OI. Following this, participants are randomised to one of the two treatment arms (TPTD/ZA or SC). Those in TPTD/ZA arm have 2 years TPTD then get an infusion of ZA and then are followed up without treatment for 3 years. Follow up in total is about 5 years.

**Are you still recruiting?**
Yes we are. If someone is interested it is possible to email the Topaz study team (topaz.trial@ed.ac.uk) and we can put you in touch with the closest centre.

**Do you have patient involvement?**
Yes, we involved patients in the study design right from the start and we have had very good support from the BBS in the UK.

**Any messages for the readers of OIFE Magazine?**
Yes! This is an important study which could change the treatment approaches in adults with OI. If you are interested in taking part and are based in UK, Ireland, Netherlands, Denmark or France please get in touch to register your interest.

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**Clinical trial: Investigating Denosumab in children**

*Interview with Dr. Jörg Oliver Semler, Paediatrician, OIFE Medical Advisory Board (MAB)*

**How did the project of investigating denosumab in children start?**
Regarding the use of denosumab in OI there are a few projects which have to be separated. At the beginning there were a few case studies and a small clinical trial which was performed in Cologne on our own initiative due to scientific interest. Some years later the pharma company Amgen initiated an international multicenter trial with the aim to approve denosumab for the treatment of children with OI.

In 2012 we realized that our usually used bisphosphonate treatment was not effective in 4 of our children. Bone resorption markers in the urine responded not in the same way as in other children. Therefore, we tried to find the reason and did genetic testing, elucidating the genetic cause of OI type VI. In this special group of patients an increased bone resorption and not an impaired production of collagen is the reason of the fragile bones. After looking more detailed into the pathophysiology we decided that the osteoclastic antibody denosumab would be a suitable treatment and might be more effective than bisphosphonates. That’s why we treated those patients with denosumab and the patients had a benefit compared to the previous bisphosphonate treatment.
Denosumab is an antibody which inactivates osteoclast directly compared to bisphosphonates which bind to the bone and get effective only after being resorbed by osteoclasts. Both agents lead to a reduced bone resorption but denosumab will vanish (degrade) after some months and can be administered directly under the skin. After we had seen that denosumab is effective in OI type 6 we did a small trial in 10 patients with “normal OI” and showed that the treatment was a bit more effective than bisphosphonates during 1 year of treatment. This trial aimed to assess the safety of denosumab and therefore the trial was not blinded or randomized. The trial was sponsored by a funding of 300,000 € by the medical faculty Cologne. The results have been published in a medical scientific journal.

What happened next?
After we had shown that denosumab was safe in 10 children with classical OI the pharma company “Amgen” decided to do a big trial to get denosumab approved in children with OI. They performed an international, multicenter, non blinded clinical trial in 130 OI-children, 2-18 years of age and they treated the children 3 years with denosumab. Because no other drug is approved in OI they had not to do a blinded or a placebo controlled trial because there is no official alternative treatment. The children received denosumab every 6 months and no bisphosphonates in addition. Primary outcome was bone mineral density measured with DXA at the lumbar spine. The trial was sponsored by the pharmaceutical company Amgen. After patients completed the 3 year study, they were given the possibility to enroll in an extension trial where treatment with denosumab will be continued. This extension trial will probably be continued till a decision about the approval of denosumab for the treatment of children with OI will be made by the FDA/EMA. Currently there are some patients who have already completed the 3 years of the “main study” and have already enrolled in the “extension study” and other patients are still treated in the main study, because they have not completed the first 3 years.

Did you have patient involvement in the project?
In our first small clinical trial the German OI society was involved in the planning of the trial. Regarding the Amgen sponsored trials (ongoing trial and extension trial) there was not much contact between the company and the patient organizations. OIF and OIFE have been informed about the trial but were not involved in the planning. But from a company’s point of view the most important questions regarding study design and treatment were raised by the authorities (FDA) and they requested to take bone density (DXA) as primary outcome parameter.

What has been the findings so far?
As mentioned above our small trial showed a higher increase of bone mineral density during one year of treatment compared to bisphosphonates. As complication we saw severe fluctuations of the calcium levels in the blood during and after treatment with denosumab. During the first few weeks after injection it is necessary to take additional calcium. In contrast at the end of the treatment period bone resorption increases and calcium is extracted from the bone and is high in blood and urine. What the clinical consequences of this fluctuation calcium levels will be has to be followed up in the future. At the moment no results of the international trial sponsored by Amgen are available because not all patients have completed the 3 years of treatment and an analysis will only be performed at the end.
And what about adults?
In adults no trials about the effect of denosumab are currently performed to my knowledge. From adults with osteoporosis we know a good effect on bone mineral density during treatment. We also know about relevant problems after pausing or ending the treatment. An increased “rebound” bone resorption has been reported, leading to new vertebral fractures. If this is the same in adults with OI and how severe this rebound is, when it will start and how it could be prevented has not been investigated at the moment. In children treated with denosumab due to other bone diseases a severe rebound has been reported in the literature.

Clinical studies in adults with OI and denosumab are definitely needed but nobody is performing these at the moment because they are highly expensive and difficult to perform. Benefits of a treatment with denosumab for adults might be an easier administration (injection under the skin versus intravenously) and the degradation after some months reducing the hypothetical risk of long term side effects.

What is the most important take home message for clinical work?
Currently we have no proof that denosumab is better than bisphosphonates and special concerns exist regarding the rebound after ending the treatment. Therefore, I cannot recommend using denosumab outside a clinical trial at the moment.

Any other messages for the readers of OIFE Magazine?
In case you or your child receive denosumab outside a clinical trial I would highly recommend checking the calcium levels in blood and in urine regularly because this parameter is influenced the most. Before results from the Amgen trial are available no additional children should be treated with denosumab, because we do not understand the effect of the drug completely. On the other side these trials are the only possibility to investigate new drugs and to improve treatment of people with OI in the future.
About the heart
As most of you will probably know, OI being a connective tissue (collagen) disorder, other parts of the body than bone can be affected. Collagen type 1 is present in different parts of the cardiovascular system, including what we call the fibrous skeleton of the heart, its valves, interventricular septum, aorta, and most other arteries, so it could be reasonable to think that collagen Type I abnormalities could also affect the heart. Although cardiovascular involvement in OI seems to be relatively rare, several cases that appear to be related to the underlying connective tissue disorder, have been reported. Most are related to abnormalities in the left-sided cardiac valves (aortic and mitral), the aortic root and ascending aorta.

Even though in most cases this is usually a progressive condition, there are also descriptions of rare cases of sudden death in asymptomatic subjects due to left ventricular rupture, aortic dissection and heart valves incompetence. Also we could think, that the reduced amount of collagen type 1 that is seen in OI could result in lower tensile strength and enlargement of both the atria and the ventricles, thus causing atrial arrhythmias and heart failure, independent of other risk factors and at an earlier age than expected. However, once again, the risk to develop this or other arrhythmias has not been evaluated.

A recent (2015) review of the literature concluded that patients with OI seem to have increased risk of heart disease compared to healthy controls. However current information about heart disorders in OI children and adults has not been systematically approached.

About the me and the Team
In the professional area, I’ve been working for 27 years as a Cardiac Physiologist at Santa Maria’s Hospital (SMH), in Lisbon. My relation and interest in OI began 21 years ago, when my younger daughter was born, with OI. Because of the lacking support, I was one of the founders of a National Association for OI in 2006 and later I got involved in OIFE. You can find our organization here: https://apoi.pt/

From the beginning APOI realized that cooperation with medical professionals was part of the process to reach our main goal of making lives of OI people better. In 2016 APOI established an internal department to help develop research studies, by helping OI people and families to come to the hospital (supporting travel and clinical costs, with volunteers help) and by helping researchers with the tasks they need (administrative, access to OI medical information, access to congresses etc.).

Cooperation at this level lead us to geneticists from SMH and start national research project to characterize the Portuguese OI population from a clinical and molecular point of view. This research was the result of an initiative of the association, and the trial design was planned with APOI, that is recognized as partner by the National Ethical Committee. Next step in this research was to move forward and “detail” the clinical characterization of OI, so we’ve invited other medical specialties to join and several “Sub-Studies” have started, one of them “OI & Heart”.
About our Study

This study aims to characterize the cardiovascular condition and evaluate the presence of potential subclinical cardiac disorders in osteogenesis imperfecta (adults and children):

- evaluate the risk of cardiovascular events on a population of OI
- correlate the mutation identified in each individual with the cardiovascular events found
- better understand the mechanisms of cardiac disorders in OI, to help doctors establish surveillance and prevention strategies, allowing patients to participate in their own health process and decision

Participants in the OI & Heart Sub-Study are invited to come to, either the Cardiology or the Paediatric Cardiology Department, in SMH (the major university hospital in the country), for 3 visits (baseline, one year and 5 years follow-up) were they have the following assessments: history and physical examination, echocardiography (with advanced features); ECG; ECG-Holter; and Ambulatory Blood Pressure Monitoring. If needed (after clinical assessment) other functional or angiographic tests may follow. Educational activities are also provided. When missing, collagen gene mutation and genetic evaluation will also be performed.

During their visits, families have the in-hospital support of an APOI volunteer and this allow us to control time so that all procedures can be scheduled or done during one single day. We have already included 66 patients. Round 50 have completed the one-year follow-up. We plan to include around 50 more patients in the coming months, but the COVID19 pandemic made us slow down recruitment.

Benefits and Impact

Several reports describe that there seems to be an increased incidence in cardiovascular events in OI but there are no recommendations regarding routine evaluation of this patients. More understanding of these problems is needed. Our goal is help to provide recommendations for routine cardiovascular follow-up of people with OI based on the characterization of cardiovascular conditions in this population.

Our aim is also that through health literacy, we can stimulate the self-management of cardiovascular risk factors and consequently increase the wellbeing, the perception of control and the overall autonomy of patients with OI. The correlation between the identified mutations and the clinical findings may also contribute to a better long-term cost-effectiveness management of the patients. The implementation of this study might also raise awareness and medical knowledge of OI among specialized health professionals.
Dreaming of a good night’s sleep and a day without caffeine
By Heidi Arponen, OIFE Medical Advisory Board

In 2015 we started a project to study fatigue, quality of sleep and its relation to daytime well-being at the University of Helsinki and Helsinki University Hospital. Initial motivation for the study arose from reports of individuals with OI on persisting daytime tiredness at international scientific OI congresses. In previously published studies, OI has been linked with fatigue i.e. lingering tiredness (Takken et al. 2004, VanBrussel et al. 2008). Research design was drawn by professors Janna Waltimo-Sirén and Outi Mäkitie. Our aim has been to explore the experience of fatigue and pain, as well as prevalence of sleep disturbances, affecting quality of living. We wanted to find out whether a medically treatable cause for the fatigue would be present. First, we conducted a questionnaire here in Finland, and then all those who were willing and able came to an overnight sleep study to look for sleep apnea. Our project was financed by Care4BrittleBones, Finnish Women Dentists’ Association, Finnish Dental Society, Sigrid Juselius Foundation, Finnish Foundation for Pediatric Research, Academy of Finland, Folkhälsan Research Foundation, and Helsinki University Hospital Research Fund.

As a result of the study, we found unexpectedly that daytime tiredness was equally experienced by the healthy controls and adults with OI. Tiredness is a subjective feeling and can be related to a number of causes. As could be expected, pain was a common problem among our 56 adults with OI. The more pain an individual had, the more exhausted they felt.

Majority of those individuals with OI and suffering from fatigue also reported of sleep disturbances. Sleep related problems were more common in OI population as compared to controls. Sleep apnea is a potentially serious disorder where breathing repeatedly pauses during sleep. If you feel tired even after a full night’s sleep, or you snore while sleeping, you might have sleep apnea. Sleep apnea is a major public health problem and can be harmful for bone health.

Following the sleep study, we found sleep apnea in as many as half of the 26 participants. Surprisingly, the presence and severity of sleep apnea did not show a connection with features usually related to it; such as severity of daytime sleepiness, anatomical narrowing of the upper airways, experienced depression symptoms, pain, or severity of OI. This fact might increase the risk of medical professionals not recognizing sleep apnea in individuals with OI, without conducting a sleep study.

Therefore, an overnight sleep study at a sleep laboratory is important in diagnosing sleep apnea. Treatment of sleep apnea include weight control, CPAP machine use during sleep, and an oral appliance that brings the lower jaw forward. Of these, the oral appliance is not suitable for persons with OI. In the future, more studies on larger participant groups would be useful in further exploring sleep in OI population.

I wish to acknowledge the help provided by the Finnish Osteogenesis Imperfecta Association in participant recruitment and Care4BrittleBones in financing the sleep studies. Together we can achieve more.

Publications:
Who are you and what is your relationship to OI?  
We are a Norwegian family of four, Anne (36), Nicolai (34), Edvard (4) and Tellef (1). Tellef was prenatally diagnosed with OI and his multiple fractures and mutation of COL1A2 gene place him in the type-III category.

How is it like to be new parents of a child with OI?  
Our main focus as parents is the same as with our oldest son, but of course there is an extra dimension to consider when we make plans and care for Tellef. The diagnosis was unfamiliar to us and we have spent a lot of time learning about OI, what to expect, how to best care for him in various situations and of course with accepting and coming to terms with the situation. We are inching closer to a “new normal” but there are always new challenges and unknowns.

Why did you decide to join the project BOOSTB4?  
OI is rare, but thankfully there is a lot of knowledge about the diagnosis internationally and with the doctors that care for Tellef. A lot of measures are taken to improve his quality of life. He has received bisphosphonate treatment regularly since he was 3 days old and we focus extensively on physical therapy. Furthermore, he has recently had corrective osteotomies of both femurs and tibias and telescopic rods now support both legs. Still, when presented with the opportunity to participate in this stem cell treatment trial we felt that the potential benefits outweighed the potential risks.

We actually learned about the research project before Tellef was born. His grandmother had scoured the internet for information about the diagnosis and available treatments. But at the time everything was too raw and fresh for us to be able to look more than a few weeks ahead at a time. After a few months we revisited the “Boost Brittle Bones Before Birth” (BOOSTB4) project information and read up on the handful of earlier cases where stem cells were administered to OI-patients. Unfortunately, we made the faulty assumption that the research was well under way and that it probably was too late to take part.

Late last year a doctor from the “TRS National Resource Centre for Rare Disorders” reached out and informed us that she had met with Dr. Götherström and Dr. Åström from the BOOSTB4 project. She had in general terms mentioned Tellef and his situation and it became clear that he was an ideal candidate for the research project. As soon as we learned this, we reached out to Dr. Götherström and the screening process began.
**Can you tell us about the project?**

The trials are split into prenatal and postnatal groups. In addition to this, there is a control group. To participate in the postnatal group the child has to be less than a year old and has to have had a definite diagnosis of OI type 3 or severe type 4. The study runs over 10 years but is based around four stem cell infusions administered with 3-5 months in between. Blood tests are performed before each treatment to ensure that there are no reaction towards the stem cells. “The infusion itself takes around 10 minutes but the patient is monitored for 48 hours after. In our case this takes place in Stockholm, Sweden. Blood samples are taken to check that the stem cells do not trigger any unwanted response. DXA scan is performed once per year. When he had his osteotomies, bone and bone marrow samples (that would otherwise be thrown away) was sent to the BOOSTB4 team. This is optional, but we opted to share biological samples with the research project. We keep a project provided diary of his perceived wellbeing (monthly and after stem cell infusions), milestones, fractures and larger medical procedures. The doctors at home have agreed to assist with testing, sending samples, and sharing information with the BOOSTB4 team. [https://www.boostb4.eu/](https://www.boostb4.eu/)

**Did you have any second thoughts?**

Of course, we gave serious thought to the potential risks raised by the trial and we were also warned that this would increase the overall treatment intensity. Even though we have little to no free time as it is, the latter was never really an issue. The former we considered more closely. Although not generalizable, we looked at the (lack of) side effects in the old cases and weighed against the potential benefits the choice was quite clear.

**Would you recommend being part of research to other parents?**

It is difficult to give a general recommendation, but we would definitely recommend the BOOSTB4 project for those in a position to participate. Of course, there are the potential benefits directly related to receiving the stem cell infusions and the hope that the project leads to a new treatment. In addition, we have had very good interactions with the professional and friendly team of doctors and researchers involved with the project. In some respects, Tellef receive double attention as we receive check-ups and advice from both our Norwegian doctors, and the BOOSTB4 doctors.

**Do you have any messages to the readers?**

As «new parents» to a child with OI the support and information we have received from OIFE and the Norwegian group, NFOI, have been tremendously helpful. We are ever so grateful. If any parents out there are curious about the project feel free to contact us.
We are pleased to inform you that we have initiated the stem cell therapy for children with Osteogenesis imperfecta under the Indo-Swedish collaboration, funded by DBT, India and VINNOVA, Sweden at Christian Medical College, Vellore. We have treated a patient severely affected by OI with the first dose of systemic and bone infusions of fetal liver-derived allogeneic mesenchymal stem cells. The eligible children under the age of 4 years will be recruited for the study. We are looking forward to recruiting children for this clinical trial in India.

Dr. Vrisha Madhuri
http://boost2b.in/
brittlebonekids@cmcvellore.ac.in

Announcement BOOSTB4

The Swedish university Karolinska Institutet sponsors an academic multicentre clinical trial to study the safety (the primary outcome) and efficacy of the investigational drug BOOST cells (fetal mesenchymal stem cells) on fracture occurrence, growth, bone mineral density and biochemical bone turnover in children who have OI.

The BOOSTB4 trial will initially open to eligible children up to 18 months of age (changed from 12 months) who have a diagnosis of OI Type III or severe Type IV with a collagen type 1 mutation. All 15 participants will receive four doses of BOOST cells four months apart. A second trial group is also planned, in which one dose of BOOST cells will be given to 15 affected fetuses before birth, followed by three doses of BOOST cells four months apart after birth. The trial is divided into two periods where the first period runs over two years, and the second period follows the child at his/her routine OI visits over an additional eight years.

The trial will take place in four countries: Stockholm in Sweden, London in the United Kingdom, Cologne in Germany and Utrecht and Leiden in the Netherlands. Please note that only one trial site, Stockholm in Sweden, is currently open for inclusion. Participants from other European countries are welcome to join the trial and travel to Sweden, or later to London in the United Kingdom when this site is open.

For more information and a complete list of trial locations visit the website www.BOOSTB4.eu.
If you have any additional questions, please contact us via the email address BOOSTB4@clintec.ki.se.

Disclaimer:
The OIFE is not involved in the design or management of this research, and as such, is neither endorsing nor supporting this study. The mission of the OIFE is to keep the OI community informed of all relevant studies. This information is made available as a service to the OI community.
New OI-causing gene

By Dr. Oliver Semler, Paediatrician, OIFE Medical Advisory Board (MAB)

A new OI-gene has been detected by a European group of researchers from Germany, UK, The Netherlands and Spain. The new type of OI is inherited in a recessive way.

In most individuals with Osteogenesis imperfecta, the disease is inherited in a dominant way and is caused by mutations in the collagen genes COL1A1 or COL1A2. In around 20%, mutations in other genes involved in collagen biosynthesis, collagen modification and secretion, the differentiation and function of osteoblasts, and bone mineralization cause OI. Thus, the quantity and quality of collagen in the cell and in the extracellular matrix is impaired. Most of these OI types are inherited in a recessive way.

During the last few years, our consortium of researchers from Germany, UK, the Netherlands and Spain elucidated the molecular reason for OI in four families. These patients suffered from numerous fractures and showed typical radiographic signs of OI, but the genetic reason remained unknown. Three different mutations in the gene KDELR2, which was previously not known to cause OI, were identified in these patients. Finally, in a collaborative effort between clinicians, geneticists and basic researchers, the pathophysiology leading to the brittle bones in these patients was unraveled and is now published in the high-ranked medical journal “American Journal of Human Genetics” (DOI: https://doi.org/10.1016/j.ajhg.2020.09.009).

Marie’s Youth Column

When you participate in a research project as a young person with OI, many questions can arise. What kind of examinations do I have to go through? What kind of treatment will they give me? Will I get the real drug or placebo? Will I have any side effects? And most of all - is the medicine working or not? Our youth coordinator Marie recently participated in a clinical research project and we bring you some of her reflections from 2020 as this edition's youth column.

Feb 20th 2020
What’s happening with the research project I’m participating in? Several of you have asked me. Today I was in for follow-up scans and blood tests, since you have to follow what happens to the medicine after you stop giving it. And unfortunately it turns out that I won’t have the results until the entire research project is finished. So I have to be patient. In general, the results show that there is an effect of the medicine, which is amazing. Personally, I’ve never ever had such a long time period without broken bones, which is even crazier! Whether it’s luck or medicine - time will tell!

Aug 9th 2020
I got news from the research project. Yesterday I was informed about what dose I received. Out of 3 different doses, I’ve received the middle dosage. And they confirmed that the medicine has had an effect (in general)! Which is really good. They don’t go into details with what effect it has had on the individual. So I have to be
patient again. Right now they’re still following us to see if the effect is declining after treatment has stopped, which also matters. Especially since I don’t know if we have the opportunity to receive the medicine again. And from the huge difference I’ve experienced in the reduction of fractures, I hope the effect doesn’t just fizzle out again. I hope we will have the opportunity to get the medicine again in the future.

Aug 20th 2020
Tomorrow I’ll HOPEFULLY get an answer from the research project! A long-awaited response. It’s been a year since I had my last dose of medication, after getting the medicine every month for a year. I’m excited about 2 things: “Has it worked?” And if so “What are the possibilities for the future?” So far I’ve only been given bone medicine, where the effect has declined over time. So if that’s the case with this medicine too, my hope is really possible to get the medicine back eventually. Because I believe it has worked! And I’ve felt a reduction in the number of broken bones. So the big question is, has it just been just random luck or the medicine? Time will tell!

Aug 21st 2020
Unfortunately, I did not get an answer today either. But I had a really good talk with my doctor who reminded me not to push myself too hard with my studies. I need to take care and feel good while doing it. I have a lower energy level and I have to take that into account. I cannot just squeeze the lemon until it cannot be squeezed anymore and then hit rock bottom. I know it already, but I needed the reminder. Sounds familiar? You know what’s best for yourself, but you forget to actually act on it?"

Work & OI
Interview with Anni Kyroläinen, Finland

Who are you and what is your relationship to OI?
I am Anni Kyröläinen. I am 31 years old and I come from Finland. I was born in Eastern Finland and after high school I moved to Helsinki from my little little hometown. I studied political science in the university of Helsinki. I have OI type 3. Nobody in my family has OI, so all this was a small surprise to my parents who got me young. I have 5 siblings, but I’m the only one with OI. I went to all normal schools and was raised like my other siblings.

Can you tell us a little bit about your job?
After high school and during my studies, I did various office and customer service jobs. I have also done some projects in politics and elections. At the end of my studies and after graduation, I headed to public administration. I worked with the Equality Ombudsman under the ministry of justice on discrimination and communication. There I was also able to use my expertise in disability issues. After that, I worked in the city of Helsinki on employment issues. I did development work related to the employment of young people and marginalized youth. I am currently working in the Ministry of Employment and the Economy. At the Ministry, my work is about work ability and changes in working life. This year, I carried out an extensive study on structural barriers to the employment of people with disabilities in Finland.
Since 2018, I have also had my own company, Riesa Consultative, with three friends of mine. We do service design and consulting related to accessibility and equality. You can read more about us here: https://www.riesa.io/en/accessibility/

**How can we improve employment of people with OI and other disabilities?**

Services for persons with disabilities, including personal assistance and transportation services, play a big role when it comes to their ability to work and inclusion in working life. Practices vary in different municipalities, which is why attention should be paid to the developing regional equality. In addition, services should support employment better than before. For example, persons with disabilities who need transportation services are not currently entitled to reimbursement of expenses caused by travel during the working day.

Public employment services are also important for the employment of persons with disabilities. The services should be developed in collaboration with organisations and experts by experience. People working in the employment services need more information about the rights of persons with disabilities and about their services. Persons with disabilities on disability pension, who are willing to participate in working life or employment services, should be taken into account.

Also, the network of service providers for persons with disabilities is complex. It would be important that, for example with the help of work capacity coordinators, measures be developed ensuring that the services provided are timely and multidisciplinary.

One of the barriers to employment could lie in the employer’s attitude or the threshold involved in recruiting a person with a disability. Information should be disseminated about targeted training, for example. On the other hand, one challenge to employment can be the fact that persons with disabilities do not have work experience. This could be influenced through guidance for students, improved accessibility, dissemination of information, and new kind of cooperation between educational institutions and service providers for persons with disabilities. All politics should be disability politics. We are not targets of care, but citizens, schoolchildren, customers and employees like everyone else.

**What have been your greatest challenges in education and work?**

Lack of accessibility in society in general. Sometimes also attitudes. I feel I need to do more and prove more that I am taken seriously.

**What is the status of employment for people with OI in Finland and other countries?**

My feeling is that for some reason, disabled people who have OI, are more likely to be in employment when compared to other people with disabilities. I don't know why. I wish the opportunities were the same for everyone. The employment rate of people with disabilities varies from country to country. There can be many reasons, services and support systems are certainly the biggest.

**You are also on the national team of wheelchair hockey we have heard?**

I've been playing floorball for over ten years. The last two years I have been a national team player. I have loved the sport since childhood, I am a football fan and for a long time I longed for the team sport that would suit me. When I moved to Helsinki, I immediately went to training and fell in love. The sport is tactical and fast-paced. You can use your game skills on the field and you can forget about all the other things.
What are your dreams and hopes for the future?
I hope to do significant things for accessibility and equality. I hope that my work in the ministry and in our company will make accessibility mainstream and improve opportunities for people with disabilities. I also hope that the Finnish national team will win the European Championship next year.

Any messages for OIFE and/or the readers of OIFE Magazine?
Keep the issue of people with disabilities and accessibility on the agenda! We do not have to settle for less. We must demand equal rights in all aspects of life!

New OIFE Member:
Indian Osteogenesis Imperfecta Foundation (IOIF)

Interview with Archana Ravindra, President IOIF

As the president of the Indian Osteogenesis Imperfecta Foundation (IOIF), I am honoured to meet you all. I am Archana Ravindra from Bangalore, India. I was born with OI and was formally diagnosed at the age of 4 by local doctors. I have tried to lead a very normal life by attending schools, college, and was the director of a small Montessori preschool for children for 17 years.

I have been fortunate to have a family with a very broad outlook into the future which helped me to explore opportunities to contribute positively to the community.
Can you tell us about the situation for people with OI in India?
India is a country with a very large population and limited exposure and access to health-related information. Although this is changing rapidly with villages modernizing; it will take several decades before we can reach the same levels as many western countries. People living in cities are better exposed and have financial affordability for medical services. Literacy being a big game changer in educating the population of rare diseases, we are slowly combating the challenges by taking small and effective steps to bring about acceptance, toleration and social inclusion in the community.

Can you tell us about your organization?
Indian Osteogenesis Imperfecta Foundation (IOIF), a non-profit entity was founded by 3 passionate people whose goals seem to align with each other on 29th May 2018. Other than me, Dr. Prashanth Inna; a specialist in Paediatric Orthopaedics has been playing a critical role in shaping our organization. We also have on board, Dr. Abhishek Bhasme; a Paediatric Orthopaedic surgeon who has been very supportive to our organization and the cause with his skills, knowledge and time for a year now. We strongly believe that awareness and education can bring about the changes we want to see in our community.

Tell us about your members!
Since its inception in 2018, our small but growing organization has just over 40 registered members with OI. However, their family members and other non-OI participants volunteer their services during events. We have been using social media like FB, Instagram and word of mouth by orthopaedic doctors from different hospitals, youth volunteers and public events to spread the word about us.

What are the biggest challenges for your organization?
As a country with a dense population and with limited resources including literacy, fist tight economy it has been quite challenging to spread the word and add more people with OI into the registry. We
find many people with OI lack a diagnosis because of too little awareness. Raising funds to host activities and funds for medical treatments such as surgeries, or provide aids like wheelchairs, walkers etc. has been challenging. We hope for this and much more to change with bringing awareness of OI to the public.

**Do you have projects you want to tell us about?**
Our projects include forming a database of patients and caregivers across Karnataka state (our home state). We have been trying to reach out to patients, mainly by getting in touch with the government hospitals which cater to the vast majority of patients who are financially challenged. We plan to reach out to doctors in the private health sector as well. We feel that to get more patients on board, it is better to focus on the treating physicians rather than trying to reach out through the general public.

Our biggest success so far has been the celebration of Wishbone Day in 2019. It was the first time in Bangalore a community awareness effort was ever made for OI. The event was organized in collaboration with Organization for Rare Diseases India (ORDI) and it attracted nearly 200 volunteers who met and spoke about OI with the general public.

**What are your plans for the future?**
Presently, we are focusing on covering the patients who come to paediatric orthopaedic centres in Bangalore. Going forward, we plan to develop a network of volunteers across the whole of Karnataka, thereby extending our coverage to the whole state.

We intend to develop a database of all the patients with OI, starting from Karnataka and then spreading to the whole of India. This would involve all the aspects of their evaluation and treatment. We plan to utilize this information to collaborate with the partner organizations across the globe with an aim to contribute to better understanding of the disease.

We intend to make a database of all the doctors involved in the care of OI in India, and utilize this to contribute to better treatment in the form of suitable medical research on OI. At IOIF, we plan to extend the awareness and education about OI to the general public, through proper utilization of the various mass media agencies.

Presently, we are arranging funds for financing the treatments of patients as and when they contact us for assistance. We plan to develop a corpus of funds so that we have ready financial assistance available. In the near future we wish to build networking with the government agencies that would help us to make suitable policies for patients with OI. In future, we endeavour to actively work in this direction.

IOIF intends to actively reach out to other organizations, form an active consortium and take the cause of OI forward
Who are you and what do you do and in what way has OI affected your art?
I’m a multidisciplinary artist and an activist making political disability arts. My passions are structural violence, aesthetics of assistive devices and issues related to women with disabilities. My background is in textile arts but nowadays I also use hard materials and make sculpture, installation, video and performance too.

I come from a family, which has a history of four generations of visual impairments from my father’s side and a history of four generations of brittle bones disease from my mother’s side. As a female artist with two kids (8 y and 11 y, both with OI) my everyday life is typical balancing between work and parenting. When fractures occur to kids, Grandma or me, we just change routines to well-tested practices to continue life as normally as possible. Our lifestyle that physically privileged people often want to see as adaptation, extreme courage or surviving, represent for me refined and cultured disability that has passed down from one generation to another.

What projects are you currently working on?
In 2019-2021 I'm making "Empathy objects" with three years funding by Arts Promotion Centre Finland. They are portable street artworks in dialogue with the environment about thoughtfulness, unconscious needs and our dependencies on other people. For example "Poverty jacket" has a tablet on its back, screening a video about how it feels like to be a poor wheelchair user in the jungle of bureaucracy. The film screening on the back is this: [https://youtu.be/O3RfgDWPZVc](https://youtu.be/O3RfgDWPZVc)
The jacket is connected to a headpiece resembling the measurement cap of the electroencephalogram that imaginatively turns the user’s thoughts into video. In 21st century, Western and Scandinavian poverty is often not outwardly visible and cannot be inferred from clothing or phone model. I wanted to visualize the inner feelings and for humour, animals play the role of authorities.

Then in September one of my short films, "Reflector of Living Will" will be screened at Helsinki International Film Festival. It tells about Maria, living with arthritis who gets tired of the rehabilitating attitude of her new care robot and reprograms it to fulfill her true needs.

**Please tell us how you became part of the film festival about disability**
July 2020 was second time for me to take part to Easterseals Disability Film Challenge that actor Nic Novicki launched 2014 in response to seeing disabilities underrepresented both in front of and behind the camera. After spending the spring isolated, home-schooling the kids I wanted to challenge myself by making a 5 min short film in five days, from the script to final editing. As the given theme was "My Story" and due to COVID everything had to be done at home, I invited my Mom to talk about OI with my kids. The result was a colorful mix of archive photographs and fracture memories titled: "Great-GrandPa looked like E.T."

**Why do you do what you do?**
I want to do my part to change the world. Globally, even in Finland, people with disabilities often don’t understand what kind of rights we have. Discrimination produced by culture, by human perceptions and inaccessible environment runs so deep. Charity and human rights often get mixed up even in decision-making.

**What’s your scariest experience related to your work?**
In a welfare country (or what Finland used to be) I’m most of the time biting the hand that feeds me. The scariest thing is to criticize authorities and services which I am depended on in many ways. On the other hand, my experience is that most of the time, bringing up nasty things leads to better decision and understanding.
What role does the artist have in society?
For me, art is a means of communication. A visual way to tell about things that happen in the margin. As a disability artist I would like to change the perceptions of disability. I’m especially intrigued by the culture of persons with disabilities who are born with their piquant features. I am worried about, what will happen if we eliminate inborn disabilities in the future by genetic engineering and prenatal screenings. If people become disabled mostly later in life, disability culture will change.

What is your dream project?
To be curated to a well-known international exhibition. Break boundaries between disability arts and mainstream. But without giving up my crippled esthetics and disrupting themes. Like my statement which is: "Why aren’t rare diseases protected on the plea of biodiversity?"

Do you have any messages to readers?
My message is in the last sentences in the film where we are taking a cast shower on the floor with my Mom and kids. We count that 8 people out of 9 born in our family have inherited OI. The only uncle who was born with stronger bones drank himself to death before his 50th birthday. For us living with OI means strength, not weakness. My whole career is based on it.

Get in Touch!

OIFE needs Patient representatives!

Are you interested in research & policy work?
As a European umbrella organization, we are more and more often asked to recruit patient representatives for research projects related to OI. This includes both clinical research and basic research. OIFE therefore needs to recruit and educate more people who can represent the voices of people with OI & their families in ongoing and upcoming research projects (patient representatives).

Sign up to join our list of patient representatives!
We have developed a database where people who are interested can register and tell us about their experience and what kinds of research they want to be involved in. Are you interested in becoming a patient representative in research & development and/or a patient expert? Then sign up here and we will get in touch: https://forms.gle/AedtyTJ3zq3LqofR6
Who can represent OIFE as patient representatives?

The answer to this question can vary from project to project, based on what kind of competence and experience is needed. According to guidelines from EURORDIS, terms “patients” and “patient representatives” are used interchangeably and refer to people living with OI and their family members or carers as well as people from organisations that represent the interests of people with OI (i.e. from patient organisations or patient groups). The term do not include people who have a professional role in health and social care services.

We are searching experts in Women’s Health & OI

Are you working with women’s health issues (hormones, menopause, pregnancy, gynaecology etc)? Or do you know any doctors or other professionals in Europe and beyond who have more than average knowledge about this topic? Or who would like to become a resource person in this field? Then the OIFE would like to get in touch with you! Please contact us on office@oife.org

OI TeleECHO Clinic Series

The Osteogenesis Imperfecta Foundation in the US recently announced the educational program OI TeleECHO Clinic Series. The goal is to build capacity to safely and effectively diagnose and treat osteogenesis imperfecta (OI). The program is a partnership with Project ECHO™ (Extension for Community Healthcare Outcomes).

This series uses Zoom videoconferencing, and will take place on second Wednesday of every month at 3pm EST (9pm CET). In each monthly session, faculty members or guest speakers will present a brief didactic presentation, followed by participant-led case presentations and group discussion of the presented cases. The faculty encourages participants to present case studies related to OI at each session. Recordings of each didactic presentation are posted after each session.

The interdisciplinary faculty of the OI ECHO includes:

- Frank Rauch, MD (Chair)
  Shriners Hospital for Children – Canada
  Professor of Pediatrics, McGill University Montreal, Qc, Canada
- Jeanne M. Franzone, MD
  Pediatric Orthopaedic Surgeon
  Nemours/A.I. duPont Hospital for Children – Wilmington, DE
- Sandesh C.S. Nagamani, MD
  Associate Professor, Department of Molecular and Human Genetics and Internal Medicine, Baylor College of Medicine, Houston, TX

A flyer with the full schedule can be found here: https://bit.ly/2TQaFzx

All the sessions have been added to the OIFE Event Calendar.

If you have any questions, please contact Michael Stewart at Mstewart@oif.org
The International Children's Bone Health Conference (ICCBH) is hosting a virtual forum from November 18-20 and we encourage professionals and patient representatives to attend.

The meeting programme of ICCBH Virtual Forum is designed for healthcare professionals and researchers. Pharma industry regulations do not allow open invitations to members of the public to attend. However, patient organisation representatives are encouraged to register to attend the meeting provided they meet the following criteria:

- They represent a formally registered international organisation with elections (or national with some international activities) and
- They are a member of the management group or are officially nominated by the management group or are a paid member of staff.

On November 18th, OIFE & OIF will together moderate a workshop on patient involvement in research.

**Workshop Nov 18th 17.30 – 18.30 CET: Patient involvement in research**
Moderators: Tracy Hart (OIF)/Ingunn Westerheim (OIFE)

Short guide to patient partnerships in rare disease research projects  
**Virginie Bros-Facer** (Paris, France)

What I learned from involving patients in my research  
**Lars Folkestad** (Odense, Denmark)

Patient involvement – A box to tick off or making a real difference?  
**Ingunn Westerheim** (Oslo, Norway)

NEW AFFILIATED PARTNERS ERN BOND
On August 31st the inclusion of Affiliated Partners to ERN BOND was finalized. 16 centres from 10 countries are now part of ERN BOND as affiliated partners (not full members). Their expertise will support all the network’s activities and will facilitate the sharing of knowledge across the EU. The process to include more full members in ERN BOND is delayed due to Covid-19 and final clarification from EU to the pending applications is expected earliest January 2021. Read more about the difference between full and affiliated members in ERN BOND on our webpage: [https://bit.ly/37o9zTE](https://bit.ly/37o9zTE)

HOW CAN ERNs BE USEFUL TO PATIENTS?
How can we create awareness about ERNs and implement ERNs as a natural part of the patient journey of the healthcare systems in different European countries? OIFE is working actively together with ERN BOND and EURORDIS to solve these challenges and make the ERNs stronger and more known in Europe. See the new animation film (available in multiple languages) with the EU Commission’s vision on how the ERNs can support patients with rare diseases: [https://bit.ly/34eTzSb](https://bit.ly/34eTzSb)

„BONE REPORTS“ - NEW PUBLICATION
Bone reports is published by the European Calsified Tissue Society (ECTS). It is an interdisciplinary forum for the rapid publication of Original Research Articles and Case Reports across basic, translational and clinical aspects of bone and mineral metabolism. Bone report is a companion title to BONE and an online only, open access, peer reviewed journal with a CiteScore of 3.4.

News in Brief
COVID-19 HELPLINE IN ORPHANET JOURNAL
ERN BOND created the “COVID-19 Helpline for Rare Bone Diseases”, in collaboration with Italian experts and Patient Associations including the Italian OI-organization ASITOI. The Orphanet Journal has now published an article about the successful collaborative initiative. Read more about the article and an interview with ASITOI’s Leonardo Panzeri on the OIFE webpage: https://bit.ly/2TbSDaY

EURORDIS SUMMER SCHOOL
Did you know that four people from OIFE has graduated from EURORDIS Summer School already? It has given us more knowledge about patient involvement and medicine research and development. Are you interested in becoming the fifth so-called patient expert from OIFE? Then apply for Summer School from EURORDIS before November 15th: https://bit.ly/34gQpgO

OI & DENTAL ISSUES
OI was on the cover on the July issue in the magazine of the American Dental Association (ADA). The cover story, "Malocclusion Traits and Oral Health-Related Quality of Life in Children With Osteogenesis Imperfecta: A Cross-Sectional Study," looked at the self-reported responses of 138 children and adolescents with OI to the Child Perceptions Questionnaire, a questionnaire developed to measure the general oral health-related quality of life of children. Among other findings the study also found adolescents reported malocclusion had more of an effect on function and oral symptoms than on emotional and social well-being. https://bit.ly/3jhMjZX

FASSIER-DUVAL RODS 20 YEARS!
Did you know that the Fassier-Duval Rod is 20 years? Dr. Fassier from Shriners Hospitals for Children Canada originally came up with the idea of an extendable surgical rod that could be inserted without having to open joints and could expand with the child as they grew.

OI-ACTIVIST IN VOGUE
Shani Dhanda from the UK is on a mission to make the world more inclusive for disabled people. In this Vogue interview she speaks about the importance of universal design and how the pandemic could prompt a paradigm shift in disabled people’s working lives. We are proud of you Shani! Read the Vogue interview here: https://bit.ly/2HncVLG
OI IN NEW ZEALAND
Umi Asaka was born into activism. Both she and her mother Yuho have OI and faced significant discrimination in Japan. Following in her mother’s activist footsteps, Umi is now finishing a degree in social work at the University of Otago in New Zealand.

She is passionate about improving the rights of society’s most vulnerable and is working to create change for young people and those with disabilities. With lived experience of disability and strong beliefs in social justice, she is driven and inspired to make an impact on the world. Check out this documentary about their experiences: https://youtu.be/xqaufVBHTXo

FACT SHEET ON CLINICAL TRIALS
The OIF has created a new factsheet called "What You Need to Know About Clinical Trials" about the importance of clinical trials and how people with OI can get involved in helping expand medical knowledge: https://bit.ly/3okI3N8

In addition to the database mentioned in the factsheet there is another one with European trials: https://www.clinicaltrialsregister.eu/

OI IN KENYA
There is no formal OI-organization in Kenya, Africa at the moment. But there is movement and a WhatsApp-group has been created to gather people who would be interested in being part of an OI-group at a later stage. Are you in contact with people with OI from Kenya? Then tell them about the link to the WhatsApp-group: https://bit.ly/3obRA9a

HEART 2 HEART HELPING KIDS WITH OI
Have you heard about the charity Heart 2 Heart OI Kids? It’s a grass roots charity run by Mary Peterson Suri, with the goal of matching medical equipment from OI kids in the US to OI kids in developing countries. Below is a photo of the excited kids in the Philippines and Indonesia waiting for mobility aids and equipment donated from families in the US. Another shipment has been made to kids in Ghana, Nigeria and Ecuador. What would the world be like without a little help from friends around the world? Read more and get in touch to help here: https://www.facebook.com/OIKheart2heart/
CALL FOR ABSTRACTS!

OIF Virtual Young Investigators Symposium

Basic, Translational and Clinical Aspects of Osteogenesis Imperfecta and Other Rare Bone Disorders

The Osteogenesis Imperfecta Foundation (OIF) is seeking abstract submissions from young investigators working in the field of osteogenesis imperfecta (OI) and/or rare bone disease research to be presented at the 2020 OIF Virtual Young Investigator Symposium on Tuesday, November 17 from 5pm-8pm EST.

This virtual one-day meeting is being offered as a pre-meeting event for the 2020 International Conference on Children’s Bone Health’s Virtual Forum and is open to US and international participants. Investigators are welcome to submit abstracts to both meetings. Submissions for the Young Investigator Symposium will be reviewed by a scientific review committee and selected abstracts will be invited to present a 10-15 minute overview of their work with 5 minutes for questions from the audience. Investigators are encouraged to share novel findings.

For more information about the ICCBH Virtual Forum visit www.iccbh.org

Eligibility

- Must be a medical or graduate student
  OR post-doctoral fellow
  OR young faculty within 10 years of last graduate degree

- Areas of research include, but are not limited to: Osteogenesis imperfecta, Paget’s disease of bone, Generalized Arterial Calcification of Infancy (GACI), Hypophosphatemia, Autosomal Recessive Hypophosphatemic Rickets (ARHR2), Osteopetrosis, Fibrodysplasia Ossificans Progressiva (FOP), Fibrous Dysplasia and McCune Albright Syndrome, Hypophosphatasia (HPP), Gorham-Stout Disease, Melerheostosis, Jansen’s disease, X-Linked Hypophosphatemia (XLH), Multiple Hereditary Exostoses (MHE), and Multicentric Carpalarsal Osteolysis Syndrome (MCTO).

The OI Foundation is committed to supporting research to help improve the quality of life for people living with OI. This often times includes engaging groups throughout the rare bone disease community, such as the Rare Bone Disease Alliance (RBDA www.rbdaalliance.org). The OI Foundation is pleased to open this meeting to the entire rare bone disease research community.

Date of Meeting: November 17, 2020 from 5pm-8pm EST
Deadline for Submission: October 1, 2020
To Apply: Visit www.oif.org/symposium or contact Erika Carter at ecarter@oif.org
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— Dr Green, HSS, NY

"I am extremely grateful to Dr. Fassier and to the developers of this system for allowing me to provide improved care and quality of life to my patients"
— Dr Esposito, Omaha, NE

"In conjunction with improved medical treatments, the FD-rod helped to significantly increase the quality of life of OI patients of all disease severity levels"
— Dr Wirth, Stuttgart, DE

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GUIDE ON PATIENT PARTNERSHIPS

The European Joint Programme on Rare Diseases "Short guide on patient partnerships in rare diseases research projects" aims to encourage fruitful, sustainable and enduring partnerships between scientists and patient organisations, co-leading the way for systematic patient-centered research. Providing definition, examples, testimonials of patient partnerships, describing its benefits, preventing common pitfalls and accompanying applicants, this guide will support applicants to describe the role and added value of patient partnerships in research proposals. Download the guide here: https://bit.ly/3mObGVk

OIFE Calendar

For an updated list of events & conferences - see OIFE's web calendar:

Contact

Ingunn Westerheim (President): president@oife.org
Ute Wallentin (Coord. Social Network): socialnetwork@oife.org
Stefanie Wagner (newsletter editor and secretary): secretary@oife.org
Stephanie Claeyns and Marie Holm Laursen (Youth Coordinators): youth@oife.org

Website: http://www.oife.org
Facebook: www.facebook.com/OIFEPAGE
Twitter: @OIFE_OI