Editorial
By Rebecca Tvedt Skarberg, ePag ERN BOND

Keynote speech from Rare 2030: “It has been a great honour to be a part of Rare2030. Building tomorrow’s roadmap for rare diseases is a project close to my heart, simply because I myself recognize the obstacles of rarity just all too well. I remember being in Romania in 2019 at the first workshop, thinking that 2030 seemed to be lightyears away. We who live with medical conditions feel that change and progress is too slow in the rare disease field. It seems that many of us are still facing the same obstacles as our forerunners did 30 years back. It’s a long road to a diagnosis, an exhausting search for information that might not be out there, research and treatment options that might not exist or not exist in your corner of the world or even worse; are too expensive to access. Enough is enough! We need to change this. Within this two-year project of Rare2030 we were ironically reminded of how quickly things can change. In 2020 our workshops had to be moved online, and we all know why.”
Thanks to the Council’s recommendations of 2009 we have made some big steps forward. We have national plans in many countries. We are more connected through the ERNs (European Reference Networks). In some areas we have new treatment and a faster diagnosis. And I dare say our voices are growing stronger in parallel with these breakthroughs. Look where we’re at!

But, my friends, there is no time to slow down. The clock is ticking, people are waiting for a chance to fulfil their potentials, a chance for independence. People are still waiting for equal rights to a precise diagnosis, to the newest innovations, to involvement and care on their own terms. Quite simply an equal right to feel safe in the hands of healthcare. We have to keep moving. Because rarity is not a problem to fix or overcome once and for all. Rarity impacts our lives on top of the condition itself. Rarity will always play a role in the choices we make on how we organize healthcare. Let’s keep working on making the impact of rarity as manageable as possible.

The recommendations of Rare2030 show us that we still need a protecting hand over rare diseases when priorities are set, when budgets are debated and when healthcare becomes a tug of war. If we handle rarity disease by disease, we will never reach enough prevalence, documentation or profit to compete with other just causes. This will not lead us to the preferred scenario in Rare2030 of a society of social justice.

We need a new policy framework for rare disease at EU level. The ERNs prove that great progress can be made when we work together. But these infrastructures are not enough. With the suggested new policy framework, we can speed up the diagnosis process for the many ultra rare diseases. We can continue to find treatments instead of putting band aids on symptoms. We can share data more efficiently and make research driven by needs. Most importantly, the expertise will have a chance to trickle down to local services so that we make sure to leave no one behind.

We can stop expecting parents to become scientists, doctors and coordinators in their free time. Having a rare condition should not have to be a fulltime job. We have our own lives to live, we expect our voices to continue to be listened to. In 2030 our healthcare will not be left to chance.

I have high hopes for where the recommendations of Rare2030 will lead us. The pandemic has shown us that we have the ability to work together and the infrastructure to develop a vaccine in under one year. And on that note let’s continue fuelling the field of rare diseases. Let’s continue pushing for change and working together on the road to 2030!”
What is the OIFE doing?

It’s been a while since the last magazine, but there have been other activities taking time and energy. And not to forget – giving us a lot of attention! But before I go to the positive news, I’d like to take a moment to pay a tribute to the three important people who passed away in 2020: First Gemma Geisman, who founded the OI Foundation 50 years ago, then Peter Radtke who founded the German OI-organization and last but not least Margaret Grant, who founded the Brittle Bone Society as the first OI-organization in the world. May you all rest in peace. The OI-community will be forever grateful for what you all started.

Then to the positive news. During the whole month of February Anna Rossi’s well planned awareness campaign on living with OI, was building up to Rare Disease Day - a day that usually creates a lot of attention to rare conditions. But this year became special. Rare Disease Day was expanded to Rare Disease Week - packed with meetings, events, politics and positive attention for the OI-community. Read all about it in a special section in this magazine. We will try to use some of the attention created to increase the visibility of OIFE and our member organization’s work in the long run.

Meetings and events

Videocalls & webinars the last months have included many meetings related to The IMPACT survey, The Photo Voice Project (#RareBoneMobility) and the EuRR-Bone OI Working group. Ingunn has also attended monthly OI TeleECHO webinars hosted by the OIF. There have also been videocalls (VCs) with OIFE members, volunteers and individual members of OIFE MAB in addition to:

- EC-meetings Nov 17th, Dec 15th, Jan 19th and March 2nd
- Steering Committee IMPACT survey Nov 17th (IW and TvW)
- Launch of Rare Impact report Nov 23rd (IW)
- VCs with European Huntington Association (EHA) Dec 1st, Jan 19th & Feb 9th (IW)
- EuRR-Bone meeting for patient organizations Dec 4th (IW)
- VCs with XLH Alliance (A stronger BOND between us) Nov 9th, Dec 14th, Jan 11th & Feb 8th
- Margaret Grant Memorial Dec 18th (IW and UW)
- AMGEN & UCB, Jan 4th (IW and BVD)
- Kyowa Kirin International Jan 12th (IW and BVD)
- Workshop on OI Variant database (IW, DL, Oliver Semler and Lida Zhytnik)
- EJP RD-meetings, Jan 15th and Jan 29th (IW)
- Nordic collaboration on follow-up routines for OI (Lars Folkestad), Feb 8th (IW)
- Argerie Tsimikalis (Shriners Montreal), Feb 10th (IW)
- ICCBH Strategy stakeholders, Feb 18th (IW)
- Lecture about OI & OIFE at Mereo Biopharma staff meeting, Feb 25th (IW)
- VC on OI Variant database, March 4th (IW)
- Kick off #RareBoneMobility, March 10th (several people from OIFE)
- Council of Federations WS hosted by OIFE and EHA, March 10th (IW)
- Webinar Towards a European Health Union, March 11th (IW)
- Meeting with Wickenstones, Mereo & OIF on IMPACT, March 11th (IW and AR)
- Establishment of Norwegian quality registry of RBDs, March 12th (IW and RTS)

All the events connected to Rare Disease Week are covered in another article.
SUCCESSFUL WORKSHOP ON PATIENT INVOLVEMENT AT ICCBH
On November 18th Ingunn Westerheim, OIFE & Tracy Hart (OIF) together hosted a workshop about patient involvement at the International Conference on Children’s Bone Health (ICCBH) Virtual Forum. The workshop had more than 50 participants from Europe, US, Canada and beyond. It included 3 talks about patient involvement from different perspectives as well as a constructive Q&A and discussion with active participation from OI-experts, clinicians, researchers and patient representatives. We have received a lot of positive feedback after the event. There were several people from OIFE attending the whole conference, that brought interesting talks and very constructive workshops with input from researchers, clinicians and patient representatives from the OI and rare bone community. You can watch the workshop here: [https://youtu.be/oZ7P-zT9UJ](https://youtu.be/oZ7P-zT9UJ)

EuRR-BONE MODULE
The OI-working group (Vertical theme 3) led by dr. Wolfgang Högler from Austria has over the last 6 months had several meetings to create a proposal for clinician reported outcomes (CROMS) and patient reported outcomes (CROMS) for children and adults in the new EuRR-Bone registry for rare bone conditions. OIFE (incl. DOIG) was represented by four patient representatives in the working group: Taco van Welzenis, Rebecca T. Skarberg, Ingunn Westerheim and Claudia Finis. We used input from our Adult Health Facebook-group to have input from the larger community during the process. Read more about EuRR-Bone here: [https://eurr-bone.com](https://eurr-bone.com)

WE HAVE REACHED 1000 IN OUR PEER GROUP!
We have now reached the great milestone of one thousand members with OI in our Facebook-group "OI & Adult Health" (English language). But there is room for more people, so keep telling your OI-friends about it by sharing this link: [https://www.facebook.com/groups/OIadulthealth](https://www.facebook.com/groups/OIadulthealth)
OIFE AT EMA MEETING ON ORPHAN DRUGS
On November 30th OIFE was represented at a workshop about Orphan Drugs hosted by the European Medicines Agency (EMA). Orphan drugs are medicinal products intended for diagnosis, prevention or treatment of rare diseases. There are currently two designated orphan drugs being investigated in OI. The MA workshop is now available on YouTube: https://bit.ly/3rGMq6n

OIFE AT COUNCIL OF FEDERATIONS MEETING
One or two times a year, members of OIFE’s Executive Committee attend the Council of Federations in EURORDIS. In November Ingunn and Bruno attended the Zoom-meeting, where more than 90 people from all over Europe took part. Topics included Rare Disease Week 2021, ERNs, Access to therapies, EU policy work, newborn screening and a lot more. It gave us valuable insights and ideas that will help us in our future OIFE work.

FIRST OIFE LEADERSHIP MEETING
February 1st the European member organizations of OIFE came together to learn about what’s happening in pharmacological research and how the patient organizations in Europe can work nationally and on a European level to improve access to new and potential treatments for OI. In addition to the delegates, leaders from the European OI-organizations were invited. Even if a treatment is approved by the EU Commission, the road might be a long and winding one before the new medicines end up in pharmacies or hospitals in the various European countries. It’s important that the national organizations have knowledge on the topic. Read more about what we can do together in the article about the IMPACT survey. Introductions included talks from Simone Boselli (EURORDIS), OIFE MAB and Oliver Gardiner and Tenna Toft from the XLH Alliance.
Rare Disease Week 2021 - “OI Takeover”

The whole month of February, Anna’s well planned awareness campaign on living with OI, was building up to Rare Disease Day. A day that usually creates a lot of attention to rare conditions and the goals we are working for. And to make it even more special, one of the faces of the EURORDIS campaign 2021 was Jon Kristian (12), a boy with OI from Norway.

In 2021 EURORDIS launched their first Rare Disease Week after inspiration from the Americans. And what a week it was! At some point I felt we had some sort of OI take over. February 22nd kicked off Rare Disease Week, where OIFE had three young patient advocates participating, meeting Members of the EU-Parliament (MEPs) after months of advocacy training. On the 23rd the Rare2030 recommendations were launched at a huge online conference, where Rebecca Tvedt Skarberg (ERN BOND ePAG) gave one of the keynotes speeches. You can read her speech as this edition’s editorial.

But it didn’t stop there. On the 24th it was time for Black Pearl Awards, where Rebecca and Ingunn won the Volunteer Award of 2021. And the photo of our Nepal friend Khim Lamichhane Kazi won the 2nd price in the photo contest. This created a lot of buzz and attention, giving us access to talk about OI on live Television on the very popular, Morning Show in Norway, and four interviews in local and national press. OIFE used the attention well and got a lot of buzz on our website and social media.

Highlights from the rare 2030 conference

On February 23rd we were at least six people from the OIFE-community represented at the big Rare 2030 conference brilliantly hosted by EURORDIS. The conference had more than 650 participants attending live (online) and even more people following on streaming. The main purpose was to present the recommendations from the Rare2030 foresight study.

Different experts presented the conclusions of the Rare 2030 Foresight Study, initiated by the European Parliament and co-funded by the European Commission Pilot Project and Preparatory Actions Programme. This two-year study with over 250 experts from across the rare disease community, has resulted in eight overarching recommendations to ensure that the future of 30 million people living with a rare disease is not left to luck or chance. Rebecca Tvedt Skarberg has represented OI in this important and time-consuming project.
Rare2030 sets out the need for a new European policy framework for rare diseases to:

- Guide the implementation of national plans for rare diseases with the same measurable objectives.
- Bring together a refreshed concerted strategy across research, digital, healthcare, social welfare complementing existing legislation
- Encourage continued investment in the field of rare diseases at both the European and national levels to ensure we do not lose momentum.

The eight final recommendations set out the roadmap for the next decade of rare disease policies.

**Recommendation 1:**
A European policy framework guiding the implementation of consistent national plans and strategies, monitored and assessed by a multistakeholder body on a regular basis.

**Recommendation 2:**
Earlier, faster and more accurate diagnosis of rare diseases through better and more consistent use of harmonised standards and programmes across Europe, new technologies and innovative approaches driven by patient-needs.

**Recommendation 3:**
A highly specialised healthcare ecosystem, with political, financial and technical support at European and national levels, that leaves no person living with a rare disease in uncertainty regarding their diagnosis, care or treatment.

**Recommendation 4:**
Guarantee the integration of people living with a rare disease in societies and economies by implementing European and national actions that recognise their social rights.

**Recommendation 5:**
A culture encouraging meaningful participation, engagement and leadership of people living with a rare disease in both the public and private sectors.

**Recommendation 6:**
Rare disease research maintained as a priority - across basic, clinical, translational and social research.

**Recommendation 7:**
Data used to its maximum to improve the health and well-being of people living with a rare disease.

**Recommendation 8:**
Improve the availability, accessibility and affordability of rare disease treatments, by attracting investments, fostering innovation and collaboration across countries, to address inequalities.

[http://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf](http://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf)
Winners of the Black Pearl Award

Ingunn Westerheim and Rebecca Tvedt Skarberg from Norway are the recipients of this year’s Black Pearl Volunteer Awards, an international award that recognizes their volunteer efforts for people with osteogenesis imperfecta (OI). The award is presented by EURORDIS a European umbrella organization with more than 950 organizations, representing people with rare diseases.

Read an excerpt from an interview (written by Kristine Hovda) the Rare Disease Alliance in Norway did with Ingunn and Rebecca after the award was announced:

“How does OI connect you?
Some people have the same religion, others have the same football club. You have the same diagnosis. How does this make you similar?

“In quite a few ways,” says Ingunn.

“I know many people with OI, since I have been the leader of both the Norwegian and the international organization for many years. Fifty per cent of those on my friend list have OI themselves, have children with OI or are professionals who work with OI. There is a strong feeling of community in the OI-field both nationally and internationally. I think it’s about having shared experiences from an early age. Having grown up with a lot of hospital stays and having the same pains, wondering about the same things and being frustrated with things that don’t work or things that can be improved.

When we were 11, we weren’t that interested in talking about OI. But with similar experiences, you understand a lot of each other without having to talk about those things. That’s probably the most important background for our friendship today.

How we got involved in volunteer work
It was coincidences that led Ingunn to start organizational work; She had just finished law school and was bored and unemployed at the age of 25. Therefore, she said yes when the nomination committee asked if she could take over as chairman of the Norwegian OI Association. The plan was to sit for a year, but it ended up with 15. Then she was ready for new and greater challenges and threw herself into the position as head of the international OI organization.

“Being a leader of the OIFE has been both exciting, terrifying but first and foremost incredibly rewarding. The position has become an all-consuming hobby where I have learned a lot and become acquainted with many exciting people, not least my partner, who is from Switzerland. We met at a conference in Finland in 2014.

I’ve been able to travel a lot, which I love very much. And not least, I have been able to lead the organization at a time when we see a lot of positive development both in terms of research, knowledge and opportunities for meaningful involvement from those who have the diagnosis themselves,” says Ingunn.
Rebecca nods.

I don’t get easily drained from energy from working on organizational work. It’s also my own services and rights I’m working to improve. We were tired from being met with more questions than answers, that no one knew anything about OI, especially in adults. That’s what’s behind our engagement.

What’s it like to win this award together?
– It’s very nice, and a little unreal. The fact that we met as eleven-year-olds, and that we’ve been following each other, that we’ve had so much fun together, it’s meant a lot,” says Rebecca.

“To be honest, I thought that the Black Pearl Awards for volunteers was one of those things you got when you retired, too long and faithful service,” says Ingunn.

– And then you got it now?
– Yes, can you imagine that! I’m not going to quit quite yet…”

Read the whole interview here:
https://oife.org/2021/02/24/winners-of-black-pearl-award-2021/

2nd PLACE EURORDIS PHOTO CONTEST
"OI CAN" a photo about life with OI by Khim Lamichhane Kazi got the 2nd place in the Eurordis Photo Contest - winning a drone kit. We send our warmest congratulations to our good friends in Nepal – doing what they can to raise awareness about OI in a country where little or no knowledge about OI exists.

WATCH THE TWO AWARDS AGAIN!
Did you miss the award show where Ingunn and Rebecca won the Volunteer Award and our friends in Nepal took 2nd place in the photo contest? Then this is your chance to watch the two awards again:

Volunteer Award
https://www.youtube.com/watch?v=ViWSNUetkSI

Photo Award:
https://www.youtube.com/watch?v=Qf3ih1AWBU

REBECCA AND INGUNN ON TV2
On February 25th Ingunn and Rebecca were hosted by the Norwegian Television on the talkshow “Good morning Norway” to talk about their experiences, their friendship and their commitment to OI and Rare Diseases community.

See the clip here (only available in Norwegian):
https://www.tv2.no/v/1636034/
OIFE at Rare Disease Week
The first Brussels Rare Disease Week (RDW) in history started February 22nd with three young advocates from OIFE members:
- Stephanie Claeys (Belgium)
- Lars Nesset Romundstad (Norway)
- Marta Rodrigues (Portugal)

Rare Disease Week is a week-long series of events organised by EURORDIS which was supposed to take place in Brussels and it included a long series of preparatory webinars. It targeted rare disease patient advocates to empower them with knowledge and skills to effectively participate in advocacy activities at the European level and influence the EU decisions that have a direct impact on the lives of people living with a rare disease. Read the impressions from Marta, Lars and Stephanie below.

Marta Rodrigues, Portugal
My name is Marta Rodrigues, I am a person living with Osteogenesis Imperfecta (OI) and I am the Vice-President of the Associação Portuguesa de Osteogénese Imperfeita - APOI (Portuguese Association for Osteogenesis Imperfecta). I truly believe that a patient-centred approach is imperative in every attempt to improve the quality of life of people living with rare diseases, from decision-making to research. The Rare Disease Week (RDW) offered exactly a program that was aimed at empowering patient advocates, providing them with the skills and knowledge on how to actively promote and raise awareness for rare diseases in decision-making at the European level. That was what inspired me to get involved.

The RDW provided a series of extremely interactive preparatory webinars and educational resources in order to prepare the participants for the week-long series of events that occurred on the week of 22 February 2021 and culminated on the Rare Disease Day on 28 February. During that time, the participants were also able to network and learn a lot from each other’s experience as well.

Although it was very overwhelming and we had a lot of information and preparation to do, overall, this experience was extremely rewarding. To meet and work with patient advocates from all over Europe towards a common goal: the improvement of the quality of life of people living with rare diseases. To have learnt from such a professional and competent team like EURORDIS is. And to have had the opportunity to put that knowledge into practice and engage with MEPs and policy makers and make them understand the importance of the role of patient advocates was an experience like no other.
Alongside Dina de Sousa from the European Huntington Association and Raquel Castro from EURORDIS, we met with MEP Marisa Matias and that was precisely one of the key messages we transmitted. We also advocated for the necessity of a new set of rare disease policies that included the new technologies and innovations and the new priorities and values in Health. MEP Marisa Matias showed a lot of interest and empathy and compromised to become a member of the Network of Parliamentary Advocates, which we considered to be a great victory.

I deeply recommend all rare disease patient advocates to participate in the next edition of Rare Disease Week. We can only strive for a better life for people living with rare diseases if we fully commit to taking the next step. This program is a great opportunity either to continue to develop the needed skills or as a starting point. By empowering yourself you are empowering the whole Rare Disease Community.

**Stephanie Claeys, Belgium**

I’m Stephanie Claeys (33), living in the very beautiful Middle-Age city Bruges, in Belgium. I was born with OI type 3.

Since my childhood I’ve always been very enthusiastic about meeting others with OI. In the beginning my parents wanted to take me to the Belgian OI organization (ZOI), after some years I asked my parents to go there myself. OI-Friendships, from Belgium and abroad are very valuable in my life.

Since 5 years I’m one of the youth coordinators of OIFE and since some months I’m also board member of the Belgian OI-organisation (ZOI). I got involved in RDW because OIFE asked people who might be interested in patient advocacy. As I love the meetings with other people with OI, I also want to help to inform them how to improve their quality of life. In my professional job I’m a patient representative in the Ethics Committee I work for, and I really want to be involved in OI advocacy in the future. I was not so familiar with EU-topics, so I wanted to learn more about it.

In my job, as I work in a hospital as administrative medical expert, we had a very busy time because of COVID-19. I was not able to follow all the RDW webinars live. But everything was recorded, so I was happy to follow this during weekends.

EURORDIS really did a good job organizing RDW. The most challenging for me was that I’m not a professional in EU topics. I know that was not necessary as we got the recorded webinars, but still. I felt not always as professional as the others were. I’ve met a lot of nice people and they all had a lot of respect for the opinion of others. It really felt like we were one group, as in the EU parliament. To have a meeting in real would have been nicer, but this was a very good alternative. They really made the best out of it.

I had a very nice talk with Frédérique Ries. She is one of the founders of Rare 2030. For me and the other Belgian patient representative this was a real advantage, as we did not have to explain why Rare Disease Week is important for the Members of Parliament (MEPs). I tried to explain her that for OI, a well organized expertise center for all age groups, is something every country should have. I told her that in the Netherlands this is the case. We do struggle a lot with doctors who have no idea what OI is. She proposed to make one for the Be-Ne-Lux (Belgium, the Netherlands and Luxembourg). If we have suggestions of how she could help us with the set up, we were welcome to contact her.

It was a challenging week, a bit overwhelming. But it gave me a very good feeling to know that together we can make a difference. So, thank you OIFE for giving us the opportunity!
Hi! My name is Lars Nesset Romundstad. I am 26 years old and live just outside of the Norwegian capital in a small university town called Ås.

I'm currently enrolled in a 2-year Master's programme in Global Development, where I am privileged to dive into the fascinating and challenging world of international politics. I am born with Osteogenesis Imperfecta type 3.

I've been a youth representative in the Norwegian Association for Osteogenesis Imperfecta (NFOI) for years, and linking the national association with our European umbrella, as well as rare disease policy advocacy on the European level is undoubtfully advantageous.

I was very lucky and grateful to be accepted into the EURORDIS Rare Disease Week events, where we were offered webinars and online discussions in the months leading up the Rare Disease Week. We were invited to dive into the complicated world of EU policy making, advocacy and institutional layout, and given the opportunity to refine our skills as advocates through practice and rehearsal, as well as actually meeting a Member of the European Parliament (MEP) during the week itself.

I had the privilege to have a meeting with the Swedish MEP David Lega, where we had a fruitful conversation on the importance of highlighting rare diseases on the international level. Specificaly on what the OI-community could benefit from when pushing for rare disease policy, as well as potential hazards and complicating matters linked to creating a uniform legislative framework which is supposed to be applicable to all EU member-states.

Even though we were unfortunate to take part in such an exciting event in the midst of global pandemic, EURORDIS did nothing short of a brilliant job when planning and executing both the preparatory webinars and the rare disease week itself. Everything fully through online platforms. Not only was it tremendously informative and instructive, it was also exciting and social. And the nerves because of meeting a MEP was still present, even though the meeting took place from the confines of my own home.

I strongly recommend Rare Disease Week for anyone interested in learning more about rare disease and international politics. I was, and still am far from an expert in any of the topics, but I am still left with a satisfactory feeling of having learnt a lot, having met a lot of interesting people, and have had several memorable and pleasant experiences. Being rare means being few, and only by working across borders with a common objective can our voices be heard. That's why Rare Disease Week is not only a fun and informative event, it is also a key element in channelling the voices of people with rare diseases.
It is an exciting time in the field of research into potential new treatments for OI. For many years, bisphosphonates have been our only therapy option but, recently, several different companies have started looking at potential new treatments specifically designed for OI.

But can we be sure that these new potential therapies will be accessible for people living with OI? No, certainly not. There are many potential obstacles. And to make sure we have strategies in place to meet those future obstacles and challenges, we need to start our preparations now. And we need to work together. This is one of the reasons we have launched the IMPACT Survey.

**What is needed and why?**

Developing a new treatment for a rare condition is a long process, where every step of the way is scrutinised by regulatory authorities (EMA and FDA) to make sure that any new potential medicine is safe, as well as proven to work. Only once the regulatory authorities have reviewed the scientific data and are convinced about this, will they give the OK for a new medicine to be sold, granting what is called a “Marketing Authorisation” or a “License”. When granted, the company in question has permission to offer the new treatment in their country or region – like the USA or the EU.

But while those steps are important, it is only the first of several, because each individual country provides their own healthcare system with their own priorities and processes. To decide what they will pay for, each country have their own special national systems to evaluate each and every new treatment before they decide if they want to pay for it or not.

The countries review additional scientific data on top of the Marketing Authorisation/ License, to understand about the condition; and how good the new treatment is at addressing the impact of that disease or condition. They look at what the condition costs their healthcare system each year (ex: fractures, hospital visits, surgeries etc); and they compare the cost for the new medication with potential savings and/or healthcare benefits of the new treatment. The different national authorities might ask for completely different information than the EMA and FDA. It can even differ between regions within a country! This means we need to develop as good data as possible both internationally and nationally.

**How can the OI-community get involved?**

The first thing that we can do, is to volunteer to participate in clinical trials, to have bigger numbers and better documentation. But not everyone can do this of course. We also have a role to play when the regulatory authorities ask our opinion. Representatives from OIFE have for instance participated in formal scientific review meetings with the European Medicines Agency on several occasions.

Last but not least, we have a role to play when our national healthcare systems will evaluate the impact that a condition has on our lives as well as the healthcare systems. And this is where the IMPACT Survey comes in as a first step. The scientific name being “Living with Osteogenesis Imperfecta: understanding experiences based on community Insight & evidence.”
Since bisphosphonates have always been given off label, and have not been specifically approved for OI (except in Italy and Japan) they have never been through the process that new medicines go through. As a consequence, both the EMA/FDA as well as national agencies do not really know a lot about OI.

With the IMPACT Survey, we are aiming to get a scientifically validated and published set of data that will show to the healthcare systems the impact that OI has, on people and also on the economics of the healthcare system. This will be important because when they will evaluate any new treatment, they will need to have a “baseline” to compare the new treatment against. The more people who answer the one time survey, the stronger documentation and data we will have.

Who is doing what?
The survey will be run by Wickenstones - an experienced scientific agency who Mereo have engaged to do the work. The overall process is governed by a Steering Committee with representatives of OIFE and OIF as well as expert OI physicians from Europe and the US treating children and adults.

The data will be gathered on a totally confidential and anonymous basis, with no personal identifiable information. It will be securely stored by Wickenstones according to Data Protection Regulations. The data will then be analysed according to the plan developed by the Steering Committee, to prepare the first publication. Once the first analysis is finished, the anonymous data will be transferred to a Data Management Committee, comprised of OIFE and OIF, and Dr. Frank Rauch (chair). In agreement with OIFE and OIF, the results will also be shared with Mereo and its partners for planning potential pathways for OI-treatments being evaluated by national payer authorities. Under no circumstances will the information be used for anything without the permission of the Data Management Committee.
So what will happen, when?
The survey will be launched in summer 2021 and we will need the help of all our member organizations and the OI-community to spread the word and encourage as many people with OI (and parents) to fill in the survey. A communication package will be put together in several different languages and shared with the national OI-organizations to assist in this process.

Since IMPACT will be a global project, the survey and communication materials will be available not only in English but also in Dutch, French, German, Italian, Portuguese, Spanish and possibly some more languages. The survey will be open for 2-3 months and once it’s completed, the results will be collected on an anonymous basis and put forward for publication in a peer-reviewed, scientific journal. This means that once the data is published, it becomes “official”, and becomes something that the national healthcare systems can formally refer to.

The collected aggregated and consolidated results for each country will also be given to the national OI member associations in the countries to use in local policy work, for instance when lobbying for multidisciplinary care or adult clinics. This means, that the more people who answer the survey in a local country, the stronger the data that can be used for later policy work, will be.

A joint project with mutual benefit
Even if the survey was originally initiated by Mereo Biopharma, the OIFE and OIF fully supports the project, because we regard it as mutual beneficial. Not only can the data be used in processes connected to Mereo’s work, but it will also be helpful for other future potential treatments being developed and for the policy work in the national organizations. Several rare disease federations are doing similar projects, simply because they see that access to orphan drugs relies on bigger and better data. This is why we strongly encourage you to support the project – and make it the biggest and best OI-survey possible. Bigger and better data matters – and with your help we can develop it. The IMPACT survey will be one of the main topics at the OIFE AGM on June 19th. But don’t hesitate to ask if you should have any questions about the project before that time.
Who are you and what is your relationship with OI?
Therese Rudolfsson and I, Lars-Göran Wadén, currently share the chairmanship of the association OI Sweden. We both have our own experience of living with OI and are wheelchair users. 7-8 years ago we participated in a summer camp in the Swedish valleys in a town called Mora. There, the first seeds were planted and with a lot of support from our Nordic OI neighbours, an interim board was eventually created.

How many people with OI does Sweden have?
It's hard to say exactly, but I heard someone say that it could be about 400 people.

Can you tell us about the situation for people with OI in Sweden?
Our opinion is that parents of children with OI generally have really good support. However, when people transition from child to adult, it gets worse. Better care for adults with OI is underway, but so far, the level of knowledge among care staff is generally low for adults. There is research on the diagnosis, but we would of course have wished that there were more resources, a greater interest and more research for a better future for us with OI.

Can you tell us about your organization?
Today we have a functioning board and about 40 members. We have just applied for and been granted membership in OIFE, which we are very grateful for. About every other month we hold member meetings with different themes, we are on Facebook and we soon have a website up and running. As a patient organization, we are involved in influencing authorities and we are working on publishing our own newsletter soon.
How many members do you have? How do you recruit more members?
At present we have about 40 members. The business plan clearly states that we will work with member recruitment. So far, we have been only semi good at it. We need to create some information material about OI-Sweden and put the material in hospitals and specialist clinics for OI.

What are the biggest challenges for your organization?
The corona pandemic has been a challenge, but we are now becoming increasingly comfortable using digital tools. We have not been so active in getting information about the association spread to recruit members, but we will get better at it.

What is your biggest success?
When the National Board of Health and Welfare called and wanted to include us in a reference group about coordination of care for OI, it felt important and we realized that we could still make a difference. When someone in Gothenburg wanted to arrange an OI week for adults and children together with us, we were happy. When individuals contact SFOI with questions and we can deliver answers or guide them to others, we understand that OI-Sweden has an important function.

What are your plans for the future?
It would have been exciting to find collaborations with other organizations, become many and strong, and work more with advocacy work. It would also have been interesting to initiate a project that highlights the people behind the OI diagnosis. A project to increase understanding among others about what it is like to live with OI.

ECTS 2020 Digital Congress
by Lidiia Zhytnik, OIFE Medical Advisory Board

European Calcified Tissue Society (ECTS) is a major community for researchers and clinicians in Europe working in the field of musculoskeletal diseases. Annual ECTS Congress brings together health and science professionals to exchange latest advances, challenges and controversies in bone and calcified tissues field.

The 47th ECTS congress went virtual in 2020. Live Prime Time sessions were held from 22-24 of October. Lidiia Zhytnik on behalf of the OIFE participated in the ECTS 2020 digital congress.

As always, ECTS congress was full of latest scientific and technological updates, clinical advices and applications. Among developing treatments for OI were reported Fresolimumab (antiTGFβ) for severe and moderate OI; Setrusumab and Romozumab (antsclerostin antibodies);
mesenchymal stem cells therapy for severe OI, and last but not least pre- and postbirth transplantation of mesenchymal stem cells in kids with severe OI. So, many different therapeutic approaches are on their way!

In addition to traditional topics (e.g. bone regulators, genetic and molecular controls of bone cells), there were fields which stood out with innovative and interesting approach, like prediction of fracture risk with artificial intelligence, connections between bone strength and gut microbiota, or between bone and metabolism (fat tissue).

Although on the first sight it might seem, that the interest of the scientific and clinical communities is concentrated mainly on common musculoskeletal diseases like osteoporosis, bone cancers and osteoarthritis, various aspects of OI were highlighted by numerous works, presented during the congress. The presentations on basic research: unusual IFITM5 mutations which connected OI types V and VI, mutations in WNT1 and OI phenotype in mice, role of a new OI gene TENT5A in the disease pathogenesis, and role of mutant collagen in the cell components stress. Translational and clinical OI research highlighted new cases of OI with MBTPS2; PLS3 mutations, disturbed bone formation in patients with OI type V, fractures during pregnancy and after delivery in women with OI and health-related quality of life in OI patients. Also, a summary of a pan-European registry for Rare Bone diseases (EurRR-Bone) was presented in the congress. We would like to express our support and gratitude to all young, established and experienced researchers who work on OI studies! Abstracts of works presented during ECTS Digital Congress 2020 were published in the Bone Reports Volume 13S (2020) https://www.sciencedirect.com/journal/bone-reports/vol/13/suppl/S

We would also like to thank Dr. Oliver Semler (Children’s Hospital, University of Cologne), who is the chair of OIFE MAB, for bringing additional attention of the audience to OI by giving a wonderful summarizing talk on clinical updates in OI.

We are also very happy to see that work, done by leaders of OI research is recognized. Our heartily congratulations to Prof. Outi Mäkitie (Children’s Hospital, University of Helsinki), who has significantly contributed to the research of OI genetic defects, a recipient of the 2020 Steven Boonen Clinical Research Award.
All it took was a quick jab, and then it was over. I got my COVID-19 vaccine on a cold but sunny afternoon in January. It felt like the usual jab, but this time the jab also came with a glimpse of light and a promise of a future with more freedom. Today I share that hope with more than 200 million people worldwide and among them a rising number of people with OI. I’m careful not to get overly optimistic. But after a year with a lot of our plans and dreams put on standby, I’m hopeful that the vaccine can help us all, to once again expand our horizons and resume the life we knew before the pandemic.

As I’m writing this, I’m aware that a lot of people with OI around Europe still haven’t gotten the vaccine yet, but I’m hoping that this has changed when you all read this. In Denmark some younger people from risk groups have been prioritized among the first people and luckily, I was one of them and so was a lot of other Danish people with OI.

Joy and relief
On the morning January 12th I got a notification on my smartphone. The notification told me, that I had just received an electronic letter from the public health authorities, and the subject line of the letter was "Offer to get vaccinated against COVID-19." Rarely has a letter from a public authority caused so much joy and relief. I found an available timeslot the next day. I booked my appointment from my phone even before I got out of bed, and 24 hours later I was vaccinated.

A lot of people have been asking about the side effects, and they are different from person to person, but you have to remember that usually the thing we call side effect is just our immune systems reacting to the vaccine. I got the Pfizer-Biontech-vaccine, and I hardly felt anything besides some soreness in my arm. After the first shot I might have felt a little bit more fatigued the following day, but it was very mild. And after all it was January, and I think we all feel a bit more tired than normal at this time of the year. So, it might just have been a coincidence.

I got the second shot three weeks later, and this time I definitely felt some fatigue. It kicked in about 12 hours after the shot and lasted for about a day. But again, it was still very mild. Like if I had been out for a fun night and got to bed a bit too late. Even if you get more severe side effects it is typically just a day of feeling feverish, having headaches or chills. For some people it can be pretty unpleasant, but it is definitely better than getting covid.

A year with new opportunities
Right now, we are under lockdown in Denmark. Restaurants, bars and museums are still closed, and travel is still restricted. So, the vaccines haven’t yet given me the opportunity to do a lot of new stuff. But after a year, where I have been mostly working from home, I am now able to drop by work again, and I have started to see my friends in indoor settings. Hopefully it will also be possible to see friends abroad soon, and I’m also planning to spend the summer in the sunny hills of Tuscany. Nothing is guaranteed in the middle of a pandemic, but with the vaccine I really feel a lot more confident that all of these things will be possible. The pandemic is not over yet, but for me the first shot of the vaccine definitely felt like the beginning of the end. And if somebody ask me if they should get the vaccine? My answer would be, that it is of course your decision, but you should know that the vaccine have been proven to be safe and extremely effective.

I see it as nothing short of a scientific miracle, and therefore and therefore I’m happy that I got mine, and I hope more people with OI around the world will soon get their shot too.
Meet the OIFE delegates

Olga Korshunova, Russia

One year ago an amazing girl called Yulia was born in our family. Unfortunately, she had OI (3 types). In the first hours after the birth I contacted Elena Meshcheryakova (the founder of the "Fragile People" Foundation in Russia) on Internet. She told me about OI in details, answered all my questions, told me where to go, how to proceed and much more. From the maternity hospital we already returned with a plan of action and understanding of how to proceed. With time I became more and more immersed, learned, read and developed. Due to the fact that there is not so much information, I remember perfectly well how difficult it was at first. At some point I felt self-confidence, I realized that there are a lot of moms in Russia who like me face the same problems with which I want to share my experience and knowledge, to provide information and maybe moral support. Thus, I returned to Elena again and now I volunteer for the Foundation.

Tell us about your organization!
Information from the foundation’s website: "The Foundation works to create a system of assistance to people with Osteogenesis Imperfecta and other bone conditions in the following areas: surgery, conservative treatment, rehabilitation, psychological assistance and social support. The Foundation also carries out other activities to educate society about the condition OI."

In our team, three of us have experienced what it means to be a parent of a child with OI. The rest of us came to the foundation at the call of our hearts. Together we look for new opportunities for fragile children and adults, solve the most unexpected tasks, rejoice at the success of children and get upset by their new fractures. In the most difficult situations, our strength is trust in each other, mutual respect and support for families with OI.

What do you do outside OI-work?
My main passion and hobby now is Kundalini yoga. Yoga helps me to stay in good mood and cope with stress. My family also love to travel. My husband, eldest son and I go for snowboarding, kiting, windsurfing and cross-country skiing. For our purposes, Yulia will share our passion for sport. I hope she will be able to do surfing.

If you were the OIFE president for a week, what would you do?
A very interesting question! A week it is quite short time to make any major changes in an already well-established system. That’s why I would gather OIFE members, fund managers and their employees, doctors dealing with non profit organizations, active parents and go to some place for the whole weekend. There we could discuss current important issues, find solutions that have not been there for a long time, discuss innovations and just meet and make friends together.

After all, together we are the power! And OIFE is one big family! The remaining 5 days I would have gathered all that this weekend we all came up with together and tried to implement as much as possible one of the major goals.

What should the OI-organizations focus on?
It seems to me that it would be good to pay special attention to the development of new treatment options of OI (all kinds of gene therapies, denosumab, stem cells, etc.).

Also psychological help to parents with OI children and OI patients so they can cope with this disease. I believe that real strength is inner strength including many characteristics like willpower, positive mood, kindness, love and gratitude for life. Developing all these characteristics will help people to live happy and productive life with or without OI.
Nadja Kreisser, Switzerland

My name is Nadja Kreisser and I have OI type I. I had more problems when I was a kid, but the disease still affects me again and again. OI has shaped my life in a positive and in a negative way and has taught me to keep fighting and to enjoy my life whenever possible.

Tell us about your organization!
The Swiss Association Osteogenesis Imperfecta (SVOI) was founded in 1986 and we include ca 150 affected people, parents, partners of affected people and sponsors. The executive board consists of 7 members with a medical or paramedical background. Three members of the Management Board are directly affected. The work of the executive board is voluntary. The management of the office is paid. We have made videos about OI and the latest video has now been released in English https://youtu.be/ILMiFjgZ2vE

We are also working on developing an information brochure and we are involved in intense networking between Swiss specialists.

What do you do outside OI-work? I work as a case manager and team leader in a hospital. I enjoy time with people I love. I like swimming, cooking and reading.

If you were the OIFE president for a week, what would you do/change? I have joined the association SVOI as a member and part of the board only for a couple of months and I am deeply impressed about all the efforts of the organization for the affected people and for a helpful networking. Therefore I wouldn’t change anything.

What is the most important job of OIFE? The global networking of specialists as well as of affected people. The sharing of a database about new studies and scientific achievements.

Do you have any other messages for the readers of OIFE magazine? I thank you for your valuable work and I am looking forward meeting you personally as soon as travelling is possible again.

Breathe, Eat, Sleep & Test!

Interview with Antonella Lo Mauro & Leonardo Panzeri (As.It.OI)

In February 2019 we started the project called “EAT, BREATHE, SLEEP with OI”. The aim of the project was to try to break the dangerous vicious circle, ensued by the pathophysiology of OI, in which breathing, sleep and body composition are tightly linked, and they can all negatively affect the quality of life in OI patients. The collection of data is now over, and we are analysing the data. Unfortunately, we had to stop the collection of data earlier than planned in March 2020 because of Covid-19 lockdown. But 27 Adult OI patients were seen before and after 6 months of a tailored dietary regime. The project is co-financed between As.It.OI and Care4BrittleBones.
Tell us about the project!

Antonella: The idea popped up in September 2016, while attending the ‘Soft Tissues & Soft Issues’ event in Oslo. During a dinner, one OI participant reported his personal experience on how his quality of life had worsened in just one year only because he gained weight. He was strongly impressed by the positive impact of non-invasive nocturnal ventilation on daytime life.

Later during the same evening, Leonardo underlined that OIers are frequently recommended to lose weight, but no indications or information on the nutritional status in OI are provided. Patients and families, therefore, end up with following a “do-it-yourself” diet, sometimes eating a lot of dairy products with the wrong idea to strengthen the bones and reduce the fractures incidence.

I immediately understood that it was definitely time to study and control the nutritional status in OI patients, together with breathing (my personal field of research) and sleep. I proposed this idea to Leonardo and As.It.OI and they agreed enthusiastically and accepted to help me in this adventure. Once home I involved Ramona and her group, thanks to a previous productive collaboration we had together for another disease. Leonardo then suggested me to involve Dr. Landoni for the sleep study. The collaborators therefore are: Politecnico di Milano for the breathing part, ICANS of the University of Milan for the nutritional part and Villa Beretta Rehabilitation Center for the sleep part.

Leonardo: I saw too many adult OIers dedicating much more energy to the career, to the family and to the hobbies rather than to their body. In adulthood, the incidence of fractures is reduced and the acquired autonomies are generally enough for a sufficient independent life. For this reason, OIers tend to reduce or skip physiotherapy and/or physical activity while eating more and therefore gaining weight. Some overweight OIers try “do-it-yourself” diet with no results and they often complain about bad quality sleep and/or morning fatigue.

Ramona: In my clinical and research activity I have always been working on dietary management and nutritional evaluation of patients suffering from rare diseases and I had the pleasure to collaborate with Antonella. So she asked me to take part at a conference of As.It.OI and in that occasion I met Leonardo and all together during the gala dinner we started with a productive brainstorming about what was missing and what we could do to implement the quality of life in OIers.

What can we do to prevent obesity for people with OI?
Patients with OI tend to become frequently overweight or even obese due to their reduced height and physical inactivity. In turn, the reduced muscle mass and strength, due to limited motor skills, correlate with an increased risk of fractures and daily fatigue, thus limiting even more the daytime activity of these patients. These clinical features can have a significant impact on quality of life and are linked to psychosocial implications that are also reflected in inappropriate calory intake and poor dietary quality.
It is therefore necessary to evaluate the nutritional status and body composition of OIers to allow the development of a personalized dietary plan. We need to educate people with OI, together with families and caregivers, on what the modifiable risk factors are, such as: vitamin D supplementation, improvement of dietary quality in terms of macro and micronutrients, exposure to the sun and weight loss in case of overweight and obesity, reduction of sedentariness, also to enhance the efficacy of their pharmacological and physical therapies.

**Why is it so important to have a healthy weight?**

Reaching and maintaining a healthy weight is important for overall health and can help to prevent and control many comorbidities. Overweight and obesity in OI patients is a condition of having a high amount of extra body fat. Adipose tissue, especially in the abdomen, seems to be a risk factor for low bone mineral density and fractures. In addition, nutritional status can also affect respiratory function, often problematic in patients with OI, especially in adulthood, due to reduced muscle mass as well as postural alteration. In particular, malnutrition, both defective and excessive, can adversely affect lung function. The adverse effects of malnutrition include decreased ventilation and reduced respiratory muscle function. Therefore, nutritional assessment should be part of OI management.

**What is the patient organization’s role compared to the professionals?**

First, communication. Second, economical and organizational support. Third, relationship with experts. As.It.O.I. in Italy is the connection between scientific projects and OIers who would otherwise be alone. For "EAT, BREATHE, SLEEP with OI", As.It.O.I. informed the OIers about the project, organized a recruitment campaign to find the participants and organized the logistic of the investigations and provided a partial reimbursement.

**Do you have any messages for the readers of OIFE Magazine?**

Remember that healthy and responsible eating associated with an adequate physical activity helps reaching and maintaining a good quality of life. This is valid for everyone. OIers, therefore, should pay even more attention! Sometimes it is enough to follow some simple rules, with moderate sacrifice: no pain, no gain. The gain is absolutely worthwhile: better breathing and energy available for your daily activities. Finally remember that the nutritionist is not a judge, but a counsellor! The importance is to be informed and aware!

---

**Be at your BEST!**

**Breathe! Eat! Sleep! Test!**

**Vivi al Meglio!**

Partecipa al progetto che potrà aiutare i medici a migliorare lo stile di vita delle persone con Osteogenesi Imperfetta!

Aderisci alla campagna di screening con i nostri specialisti!

#Be@yourBEST!
Jeanette Chedda: Fighting for Diversity!

Through social media we learned that Jeanette Chedda is running for Parliament in the Netherlands. Her main cause is disability politics and fighting for diversity. The elections are coming up in just a few days.

Can you tell us a little bit about yourself?
I am Jeanette Chedda (37), I have OI type three. I am the daughter of two amazing Surinamese parents. My father died in 2017 but I still have my mother. I am the oldest with three siblings (34, 22 and 17 years old). I am the only one with OI. I am a web editor since 2010, a disability rights activist (anti-ableism), ambassador for the Care4BrittleBones Foundation lead by Dagmar Mekking and a lot of amazing ambassadors and volunteers.

How did you end up running for Parliament?
I ended up running for Parliament because I was asked. So I applied for a spot on the list and became the number four of eighteen candidates. On March 15,16 and 17, The Netherlands is going to vote for their preferred party. So in the days after we will know if we got enough votes to enter Parliament. It is very exciting and I am optimistic. You should check out our party: bij1.org.

Why should people vote for you?
The main cause I fight for is disability rights. The biggest misconception about The Netherlands is that we take good care of our disabled community. In 2016 The Netherlands ratified the Convention on the Rights of Persons with Disabilities, CRPD.
Since then, our position in society worsened on different aspects of life (education, financially, living, care etc). Different research proves this, even the Institute for Human Rights and other disability organizations agree.

I believe that representation matters. Our community has a 0,3% representation in Dutch politics. That is one of the reasons people should vote for me. Disabled voices need to be heard more. Nothing about us, without us.

**Why are role models important?**
I grew up with no role models. In 2019 I saw someone that looked like me (a woman with South Asian roots) for the first time in the media. It was Shani Dhanda from the UK. I cried. It is so important to see people who look like you in certain positions. People who you can relate to and identify with. You can’t be what you can’t see. We need to have more diversity in our parliament. And more disabled people in decision making. Because these policies affect our lives. Our current Parliament doesn’t reflect the richness of our country. The parliament right now consists of mainly white people, mostly male.

**In what ways have OI affected your political career?**
What has been most negative is the harmful prejudices people have of disabled women of colour. I have to deal with sexism, racism and ableism. That’s a heavy burden to carry. Also managing my lack of energy in combination with the harmful urge to prove myself. I try to temper it. But that’s not always possible. It has been a life long struggle. My positive outlook on live has been an asset though. That’s an OI-thing right? A hope for a better world drives me. And the amazing people I know with OI that support me, lift me up and wish me well.

**Why do you do what you do?**
To be honest I don’t think I have a choice in this. The alternative for me is doing nothing. And doing nothing is to comply with the situation. I will not comply. I will not shut up. I will not submit. We as (disabled) people deserve more than what we are given in this world.

I grew up thinking there was something wrong with me because I have OI being a short person and using a wheelchair. I was lonely and sad and depressed a lot of my life because of this. I don’t want disabled kids to go through the same. I want them to know: you are strong, you are smart, you are worthy, you are kind, you are here and you will not disappear. You deserve a spot in this society as much as a non-disabled person does. You’re worth is not defined by your ‘productivity’. You belong in this world and I see you.

Hope and the motivation to make a better world for ALL of us drives me. I have seen the beauty of good allies in this world. That is also a driver for me. Because we can’t do this alone. Allies pull up!
In which ways do you think the OI-organizations can be better at diversity and inclusion?
Interesting question. First of all. Do your homework! And I would like OI-organizations to make more room for Black OI-people, people of colour and immigrant background. Explicitly. Actively. You need to be actively anti-racist. It’s not enough to say “Everyone is welcome here!”

How can the OIFE contribute?
You can facilitate the conversation, make room for this conversation, be more intersectional. Make room for more black people and OI-people of colour. The lack of black and brown representation really hurts me. Because I feel I am part of the OI-community but at the same time I really miss the acknowledgement and conversation about of racism and unconscious bias around race within our own community.

What do you do outside politics?
I love being active physically. I have been swimming my whole childhood. Since 2012 I have been playing wheelchair hockey (floorball) which I love. And indeed boxing. Only with a punching bag. I don’t box people. Because I still have OI.

Do you have any other messages to the readers of OIFE Magazine?
Let’s support each other more, lift each other up, share our platforms to let our important voices to be heard more, especially the Black and Brown disabled voices. Let’s be good allies and acknowledge white privilege. Not sure what that means? Please do your homework!

**Artist with OI: Pablo Ostarek**

What a coincidence! Some time ago OIFE had prepared the interview below with Pablo Ostarek, an artist with OI. And now we congratulate him for winning the 1st prize in the International Rare Disease Film Festival for his short film "This is not a person"!

The film is about the Spanish woman Elisenda (45), who didn’t get to live an independent life because of overprotection.

On [www.oife.org](http://www.oife.org) you can read more about the film project and see both the film and the award show (but only until the end of March).
Who are you and what do you do?
My name is Pablo Ostarek. I am 28 years old. I try to get by with different jobs, but my aim is to be a full time film director. Although it is hard to find a way to get paid with our projects, we have to keep trying to make that happen.

In what way has OI affected your art?
I don’t really think that OI has affected my art directly. Who I am determines the art I make, and I happen to have OI. I guess there are themes and types of stories that I have been more interested in since I was a child that we could link to having OI. The time spent at the hospital or the consequences of certain fractures have definitely had an effect on me as a human being, and have resulted in certain doubts and fears that I would not dream of shying away from in any of my projects.

In our new documentary film, my colleague Miguel Rodriguez and I look into the consequences of overprotecting a person with OI. That may be the most direct link any of my work has had with OI, but then again, in our case OI is the context, or the world our characters are in, but the film as a whole is not about OI.

You have already been accepted for many film Festivals which is fantastic!
Yeah! Miguel and I have worked hard on our short film "This is not a person". We don't have much experience in the film festival circuit but It isn't going bad considering it's our first big project. Our project was selected in about 10 film festivals so far, but we are hoping to make it into a few other official selections soon.

What projects are you currently working on?
We are working on another documentary about the living situation in Spain. We are mostly interested in the difficulties young people have to face nowadays when it comes to forming a family and being able to be completely independent. The project is still in its early phases, but we are hoping to film it some time in the new year.
Then we also have a few short film ideas, and every now and then we shoot a videoclip for friends, but money is tight, and it may be a bit tough to film everything we want in the near future. We are hopeful though!

Why do you do what you do?
I have had this idea in my head for a long time. I love movies and I just kind of started on that road one day. It isn't easy and nothing ever works out perfectly, but it does bring joy often and pursuing to become a film director gives my life purpose. There is also a certain need to create. I love music, but I'm not a musician because I'm no good in making music. I love film and writing, and so far I haven't been too bad at making it, so I've decided to keep going and learning and improving.

What themes do you pursue?
I am interested in perception. I believe the way everybody looks at and perceives what surrounds them is key to making up your mind on reality. The way every individual feels different when subjected to the same stimulus is fascinating. Human emotions and feelings can enrich a character in so many ways, make them so unique. Plunging your main character into a made up context and seeing where it takes him or her is the foundation of any story. I don't know if I can say that I pursue certain themes more than others; one day I get obsessed about the story of a lonely, depressed, self-involved character and the next I can't stop thinking about a comedic, coral, mockumentary style sitcom similar to "The Office".

What's your favourite art work?
No idea. I'm not going to think too hard on this question, so I'm keeping the answer strange: the live video that you can find on YouTube of The Rolling Stones performing at the Checkerboard Lounge in Chicago in 1981 (44:23). The whole thing is just one lovely piece of historic art. It's a documentary in its own right. It's Scorsese's 1969 Woodstock documentary in a very, very small, unplanned way. But it's original and authentic and it makes me feel great every time I watch it.
Research announcements

Romosozumab Trial for OI
The company Amgen is sponsoring a multicenter clinical trial to study the safety, pharmacokinetics, and pharmacodynamics of the investigational drug romosozumab in children and adolescents who have OI. Currently the study is open to eligible children ages 5 to less than 18 years of age who have a diagnosis of OI Type I through IV. The study has sites participating in this clinical research in the following countries: Germany, Hungary, Italy, Spain, Greece and Turkey. For more information, please visit the clinical trial registry page at: https://www.amgentrials.com/study/?id=20160227

If you have any additional questions, please contact the Amgen Call Center at +1 (866) 572-6436 or send email to medinfo@amgen.com. A Customer Service Representative will be able to find a study site near you and provide you with the site study contacts who can tell you more about the clinical research study.

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.

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

Disclaimer: The OIFE is not involved in the design or management of these research studies and as such, is neither endorsing nor supporting these studies. 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. We are available to answer questions on this or any other research announcements. Please contact the OIFE at office@oife.org if you have any questions.
**eBOOK – “ATHLETES WITHOUT PERMISSION”**

Our Polish delegate Aneta is planning to write an eBook about good examples from people with OI living in Europe who are physically active in different ways. Aneta herself has OI type IV and has tried many different sports and activities: swimming, scuba diving, crossfit and climbing. The project has the support of the OIFE.

Preferably Aneta would like to interview professional athletes with OI about their triggers, obstacles and experiences. But OI-people who are doing an interesting sport or physically activity as hobby are also of interest.

The main goal of the project is to shed light on the relationships between people with OI and their environment, including factors like overprotective parents and influence from medical professionals.

Other goals include:
- Encouraging people with OI to be physically active through sports
- Creating awareness among professionals about the benefits of sports as an alternative to physiotherapy in building muscles and bone strength
- Creating awareness about the role of the OI-organizations, peer work & role models.

Would you like to help Aneta to find the right people? Or are you an athlete yourself and would like to be featured in her book? Then send an email to office@oife.org and we'll put you in touch!

**ARE YOU USING THE OI VARIANT DATABASE?**

Professor Raymond Dalgleish moved to the University of Leicester in January 1984 and set up a group to study the genetics of OI and vascular EDS. Since 1984 he has on a volunteer basis been systematically collecting all publications describing OI-causing sequence variants (mutations). At first the database was on a static webpage, but in 2008 the data for COL1A1, COL1A2 & COL3A1 were transferred to The Leiden Open Variation Database (LOVD).

Most variants in the database are from journal publications. Some data are submitted directly from laboratories by registered “submitters”. This includes central geneticists and researchers in the OI-community. Submissions are checked before being made public.
The database has been hosted using LOVD v.2.0 on servers provided free of charge by the University of Leicester. The database contains variant data for 21 recognised OI genes.

The COL1A1 and COL1A2 data have already been migrated to Leiden University but not updated apart from variants that result in EDS. The moving of the collagen genes was funded by the EDS organizations. The data about the other OI-causing genes are still left in Leicester.

Since professor Dalgleish is retiring and no longer has the capacity to do this work, he wants to move the remaining data (19 OI-genes) to Leiden. There is a need to bring the data up to date and maintain the whole database for the future. A bioinformatician is needed to carry out the tasks and guide others regarding what needs to be done subsequently. OIFE is assisting by trying to bring people who could help in this process together. We’re interested in getting in touch with people, projects or other stakeholders:

- Who are using this database and who’s work would be compromised if these data should disappear
- Who would be willing to help with moving the database to Leiden
- Who would have the necessary skills to clean the data that is not up to date
- Who would be interested in taking a leading role in curating the data for the future

Link to the database here: [https://oi.gene.le.ac.uk/home.php](https://oi.gene.le.ac.uk/home.php)

You can also make a donation to the work of saving the platform here: [https://www.research4oi.org/project/savetheoidatabase](https://www.research4oi.org/project/savetheoidatabase)

---

PROFESSIONALS WITH EXPERIENCE ON PAIN & OI

We are interested in getting in touch with professionals (PTs, OTs, psychologists, medical doctors, researchers, pain specialists etc) who have experience/knowledge about pain & pain management in OI (or related bone diseases).

We are considering a mini conference and/or project related to pain & OI and we are interested in getting in touch with people who could contribute. We also want to get in touch with people with OI who have more knowledge than the average about the topic. Please provide us with names & emails of people we could contact by sending an email to [president@oife.org](mailto:president@oife.org)
News in brief

OIFE AGM 2021 – SAVE THE DATE
The Annual General Meeting (AGM) of OIFE will take place online on June 19th. Make sure you save the date! More info to come.

OIF CONFERENCE 2021 WILL BE ONLINE
The OI Foundation have decided to make their 2021 OIF National Conference a virtual affair. The Virtual OIF Conference will be held on July 10 - 11, 2021. We’re sorry that the American OI-community is not able to meet face to face in 2021 either. But the good thing is that people from all over the world can attend the conference. Make sure to save the date. See you there? https://oif.org/virtualconference/

WHAT IS BASILAR INVAGINATION?
CRANIOCERVICAL DEFORMITY IN OSTEOGENESIS IMPERFECTA
Dr. Suken Shah

OI & BI - BASILAR INVAGINATION
Through our peer groups on Facebook, we sometimes get in touch with people with OI who have symptoms of the rare condition Basilar invagination (BI) without being aware of what it is and what they should do about it. Because there is so little awareness about this rare but serious complication, we would like to share our knowledge on it based on information from international experts on the topic: https://oife.org/2021/01/25/what-is-basilar-invagination-bi/

HELP US KNOW MORE ABOUT COVID-19 & OI!
The OI Foundation has created a second survey focused on how the COVID-19 pandemic continues to impact the OI community in the US and worldwide. The OIFE is supporting the project. We encourage as many people with OI (or parents) as possible to provide feedback here: www.surveymonkey.com/r/OIFcovidsurvey

ERN BOND EXCHANGE PROGRAMME
Knowledge sharing and stimulating collaboration between health care professionals in ERNs. That is the aim of the Exchange Programme 2020-2022, funded by the European Commission. Read more about the programme on http://ernbond.eu/

NEWS FROM RESEARCH
Researchers from the Eindhoven University of Technology (TU/e) and Radboudumc have succeeded in growing a lifelike piece of bone tissue from human stem cells. This is the first ‘organoid’ of a bone, a simplified version of the original. The research was published in the journal Advanced Functional Materials. According to the article, the cultured bone is especially suitable for testing and designing new treatments for bone diseases such as osteoporosis or osteogenesis imperfecta. Read more: https://bit.ly/3bDjB5d
ARTICLE ON REPRODUCTIVE OPTIONS
This article from Lida Zhytnik and colleagues gives a good overview of what kind of technical options are available when a person with OI is considering to have children:
https://rdcu.be/ceuhf

WE RECOMMEND - TELEEECHO RECORDINGS
The OIFE recommend you check out the OI TeleECHO talk about Treatment in Adults with OI given by dr. Sandesh C.S. Nagamani, who's a member of the OIF Medical Advisory Board. You can find the recording here:
www.youtube.com/watch?v=4k8niZPE4bg

We also recommend this lecture on medical treatment in children by dr. Frank Rauch:
www.youtube.com/watch?v=znRThRkXPgY

Professionals worldwide can attend the monthly case discussions (OI TeleECHO) for free. https://oif.org/echo/oi/

REST IN PEACE MARGARET GRANT
We were very sad to receive the news about the passing of the founder of the Brittle Bone Society, dr. Margaret Grant. We send our condolences to her family and friends from OIFE and the international OI-community.

REST IN PEACE PETER RADTKE
We were sad to learn that the founder of the German OI-organization DOIG, Mr. Peter Radtke, has passed away, 77 years old. We send our condolences from OIFE and the international OI-community to the friends and family of Mr. Radtke

VIDEO ON REHABILITATION OF OI-CHILDREN
Through the link below you can find a video of Tellef in rehabilitation after rodding surgery, with full English text. The video is made by the Norwegian Centre of Expertise TRS kompetansesenter for sjeldne diagnoser:
www.youtube.com/watch?v=HmnmxzHcjEs
EUPATI OPEN CLASSROOM
The European Patients’ Academy on Therapeutic Innovation provides scientifically reliable, objective, comprehensive information to patients and the public on the research and development process of medicines. It aims to increase the capacity of patients to be effective advocates and advisors, e.g., in clinical trials, with regulatory authorities and in ethics committees: https://learning.eupati.eu/

NEW CO-DIRECTOR OF OI-PROGRAMME
Jeanne M. Franzone, MD, pediatric orthopaedic surgeon, has been appointed Co-Director of the multidisciplinary OI Program at Nemours/ Alfred I. duPont Hospital for Children in Wilmington, DE, USA. Dr. Franzone is joining her orthopaedic partner, Dr. Richard Kruse and pediatric geneticist, Dr. Michael Bober in this role.

Dr. Franzone completed pediatric orthopaedic fellowship training at duPont Hospital for Children in 2015, followed by advanced training in limb deformity and lengthening surgery.

She returned to Nemours in 2017 to expand the OI Program, by providing expertise in complex limb deformity correction.

MAKE YOUR BIRTHDAY SPECIAL! Help us reaching our vision of children and adults with OI living active and independent lives by creating a birthday fundraiser for OIFE on Facebook!

100% of donations goes to OIFE - Facebook charges no fees. Want to try? Go to https://www.facebook.com/fund/OIFEPAGE/

CONGRATULATIONS SHANI!
Congratulations from OIFE and the international OI-Community to Shani Dhanda from UK for making it onto the prestigious BBC 100 Influential Women in The World 2020 list.
#RareBoneMobility Photo Voice research project

Are you living with a rare bone condition? Then check out the new project #RareBoneMobility! The project is supported by the organizations OIFE and ANDO Portugal, which is a national patient organization for people with rare skeletal dysplasias in Portugal.

“We want to understand the daily life mobility challenges individuals with rare bone conditions have, how they solve these challenges – and how researchers can use the information to improve people’s mobility.”

Find out more and upload your photos on www.rarebonemobility.org

OIFE Calendar

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

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 Claeys and Marie Holm Laursen (Youth Coordinators): youth@oife.org

Website: http://www.oife.org
Facebook: www.facebook.com/OIFEPAGE
Twitter: @OIFE_OI
Instagram: oioife
LinkedIn: https://www.linkedin.com/company/oife
YouTube: OifeOrg