Editorial

To have or not to have...children

I was seventeen the first time I attended an educational course about OI. It was primarily a course for adults of small children with OI, but Rebecca and I were nerdy enough to be interested in genetics, collagen and symptoms when we were 17, so we registered in any case. And it was at this course I had a memorable talk with one of the most famous geneticists in Norway of all times – dr. Heiberg. And even if this is around 100 years ago, well at least 30, I still remember it quite vividly.

After talking about this and that – the professor proclaimed: “Whatever you do in life young lady, make sure not to get pregnant!”

And I don’t know if this is the reason or not – but I have gone through life without really considering having children of my own. Perhaps because I was afraid I would not be able to care properly for a small child? Perhaps I was afraid of what a pregnancy would do to my body? Or perhaps I was afraid of having a child with OI? Not because that would be a disaster in itself, but because it would be even more difficult caring for a child who’s fracturing when you have a disability yourself. Or perhaps I didn’t really consider it because I didn’t find a man I would consider
having children with until I was over 40. And then he happened to have OI himself. Same type as I have, only a different gene. Which leaves us with the odds of 75% chance of having a child with OI.

“Fortunately” I was so old when I met the man of my life, that I was no longer even theoretically considering children. Because of scoliosis, breathing, chronic pain, and other issues – I seriously thought it would be too hard on my body. And unlike the man of my life, I have never really had the urge to produce children of my own. Which is good, because the opposite would have made it so much harder.

I’m fine with borrowing our friend’s toddlers for a short while or playing games with the older ones for a couple of hours. But it’s not sometimes I miss regularly, to be honest. I have found other passions in life; like cultural experiences, good food and wine, friends and not to forget, my very own baby that I have adopted from Ute Wallentin – the OIFE!

And for me this is enough. I completely understand that other women (and men) with OI have different opinions, priorities and dreams and passions in their lives. I support those who choose to get pregnant without worrying about having a child with OI. And I support those who consider using one of the many options that are now existing, to check or secure that the child you are carrying does not have OI. Donor eggs, PDG, NIPT and much more. This OIFE Magazine is a special edition about pregnancy and reproduction. It covers various aspects from risks connected to pregnancy when you have OI or brittle bones, parental experiences and satisfaction with genetic counseling after receiving a prenatal diagnosis of OI, young OI-people’s knowledge about inheritance and Lida’s brilliant article about OI & reproduction. Check out the illustrations in particular! I have rarely seen such a complex topic being described in such a pedagogical way.

Tomorrow I’m leaving for the ICCBH-conference to learn even more about OI! Hopefully you will be able to read about our experiences in the next OIFE Magazine! Until then I wish you all a good summer!

OIFE president, Ingunn

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

Since the last OIFE magazine, we have used our time on planning events as the OIFE AGM, OIFE Investigator Meeting, Topical Meeting in Sweden in 2023 as well as continued activities in the OI & Pain project. We have also used time and energy on fundraising for various projects and activities. The whole OIFE EC has worked closely together to finalize the implementation of OIFE2 in Belgium after our establishment was approved by Belgian authorities in April. Led by our communication manager Anna Rossi, we have also organized a successful annual Wishbone Day campaign with the hashtag #1Voice4OI, that created a lot of buzz and global engagement as usual.

MEETINGS & WEBINARS
We have had some, but not many meetings connected to different clinical trials & research projects. This includes several different meetings with the companies Ultragenyx/Mereo, to provide input from the patient perspective. We have also had some planning meetings related to the pain project and the planning of OIFE AGM and OIFE Investigator meeting.
These are some of the other virtual meetings we have attended since the last OIFE-magazine:

- OIFE EC-meetings April 3rd (expanded), April 27th and May 24th
- EURORDIS videocall about Ukraine relief app, March 30th (IW)
- Norwegian registry for rare bone conditions, March 1st (IW)
- OIF Investigator Meeting online, April 7th (IW)
- OIF Town Hall Meeting, April 8th (IW)
- Sanofi April 21st and May 3rd (IW and BVD)
- European Hemophilia Consortium April 21st (IW)
- Kurma Partners, April 26th (IW)
- XLH Alliance, May 9th (IW)
- Kick off meeting UCB & AMGEN, May 20th (IW)
- Wickenstones about poster planning, May 24th (IW)

**EURORDIS ROUND TABLE OF COMPANIES**

The EURORDIS Round Table of Companies (ERTC) took place from April 27-28. It’s primarily an event for industry representatives, but some patient experts are also invited to attend. This meeting had the overarching topic “Laying the ground for European Action on Rare Diseases: the role of industry”. Ingunn, was invited to attend, but only had the possibility to attend parts of the meeting.

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**Highlights : 1 000 Members in April 2022**

By the end of 2021, EURORDIS had **984 members in 73 countries**, 43 of which are European countries, 27 being members of the European Union. 59 new members were approved in 2021 (a 4% increase).

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**EURORDIS AGM**

The 25th AGM of the umbrella organization EURORDIS took place May 18th and Ingunn Westerheim and Rebecca Tvedt Skarberg represented the OIFE. Highlights & success stories from EURORDIS’ 25 years were discussed and plans for the future presented. The European umbrella now include more than 1000 member organizations and plays a very important role in European policy work for rare conditions. The most important thing for OIFE, was the elections this year. Our very own patient expert Rebecca Tvedt Skarberg, was elected as one of the new Board members of EURORDIS. We wish Rebecca good luck with this important position!
ABSTRACTS FOR ECRD, ICCBH, OI2022 AND MORE
Representatives from OIFE (Ingunn, Taco, Rebecca and Claudia) and OIFE MAB have been co-authors on a record number of abstracts that have been submitted to various international conferences, which include the ECRD-conference, the ICCBH conference and OI2022. This includes several abstracts with results from the IMPACT survey, including one about the role the patient organizations played in the recruitment process of the survey, where OIFE (Ingunn) is the main author. The abstract from OIFE has been accepted both to the ECRD, ICCBH and the OI2022 conference. A big thank you to Dr. Frank Rauch, who provided assistance on the writing process! Several different representatives from OIFE have also been co-authors on abstracts about EuRR-Bone and about the OI Variant database. We are looking forward to an exciting summer and autumn, where a lot of new research about OI will be presented in various conferences in Europe.

PAIN & OI PROJECT
We hosted our second and third OI & Pain workshop, on April 26th and May 31st with more than 15 participants from US, Canada, Europe and Australia. Both workshops had assessment and measuring of pain in clinical trials and the clinic as a topic. The workshops brought interesting discussions, but we need several meetings to be able to make any recommendations on the topic. However – we established a subgroup to work on a toolbox about Pain & OI.

OIFE ANNUAL GENERAL MEETING
On June 4th we were ca. 35 people representing 18 member organizations, who came together for OIFE’s Annual General Meeting (AGM) 2022 on Zoom. The general assembly unanimously voted to approve the annual and financial reports for 2021. The proposal from OIFE’s Executive Committee to dissolve OIFE as a non profit in the Netherlands was also approved, which means we can finalize the process of moving our umbrella organization to Belgium. We’re now working to implement our new bank and to plan the first general assembly of “OIFE2” under Belgian law, which will take place on Zoom September 18th 2022. In addition to formal business, the organizations also got to learn about exciting future events. This include next year’s AGM & topical meeting in Stockholm, which will be our first face to face meeting in a long time.

What we do right now
• Our most important activities at the moment include:
  • OI & Pain project
  • Access to treatments
  • Gathering patient experience data (PED) – incl. IMPACT survey
  • Educating ourselves and our members about regulatory affairs
  • Patient engagement in R&D – both clinical trials and regulatory
  • Planning events incl.
    • 1st OIFE AGM for OIFE 2 (under Belgian law)
    • OIFE Youth Event in Poland
    • OIFE Investigator Meeting 2022
    • Topical Meeting 2023
  • Representation at conferences – ECRD, ICCBH & OI2022
  • Keep you up to date about what’s going on!
• We represent our members in many different European forums

OIFE EDUCATIONAL WEBINAR
The second part of OIFE’s annual meeting on June 4th 2022 was an educational webinar open to anyone. And a diverse group of ca 80 participants including OIFE-people, clinicians, researchers, industry people and other interested individuals came together to hear about news from the international OI-community.
**News from OIFE, ERN BOND & others**

The first part of the educational webinar included news from OIFE’s projects and activities and was presented by OIFE president Ingunn. The 2nd session included a talk from coordinator Luca Sangiorgi about how patients and doctors can access expert advice from the European reference network for rare bone disorders (ERN BOND). It also included a talk from Dutch OI-researcher Marelise Eekhoff on what a clinical trial is, why people with OI should consider taking part and what they should consider before doing so.

**News from research & development**

Marelise’s talk was a good introduction to the following sessions, which included updates from pharmaceutical research on OI. Ingunn talked about the value of patient experience data (PED) and real world data (RWD), which is the background for our biggest project in 2021 – the IMPACT survey. Arun Mistry gave updates on the results that are gradually coming out of the IMPACT survey, which include both posters and talks at various conferences as well as four scientific articles.

OIFE has also created their own abstract about how the organizations worked together to recruit more than 2200 responses for the IMPACT survey. The last part of the research session was updates from various clinical trials, including:

- The setrusumab project (Mereo & Ultragenyx)
- The romosozumab project (UCB & AMGEN)
- The BoostB4 trial (Boost Pharma)
- The Topaz-trial

**What do the delegates want?**

The 2nd part of OIFE AGM part 2 was an interactive discussion with the delegates & community members about what kind of events and topics they want to see more of in the future. The delegates provided feedback that they wanted a mix of topics including organizational development and news from OIFE projects and research. Our associate members from Chile and Peru also stressed the importance of continuing with online events, because it has great value for those who cannot travel to Europe for meetings.
**OIFE Youth Event – Our trip to Bruges**  
*Written by Simey Truong, OIFE Youth Coordinator*

OIFE Youth Event 2022 (part 1) took place in the Belgian city Bruges (Brügge) from May 5th to 8th after the last two events were cancelled due to the pandemic. The event was organized by the OIFE Youth Coordinators and the Belgian OI-organization ZOI. And how did it go? Read all about it in this report from OIFE Youth Coordinator Simey.

Finally together again!
May 5th 2022! I had been looking forward to this date for so long. After what seemed like an interminable two years, the OIFE Youth Event could finally take place again. Since Covid it had to be postponed or cancelled again and again. And many of my friends from all over Europe have been sorely missed since then. So now we were all going to meet again from 5 to 8 May in Bruges, Belgium.

Although I have been to many youth events, Bruges was very special for me. It was my first event as “Youth Coordinator” and therefore also as backup support for Stephanie, who was the host and organiser for this year. She had made it her mission to make this event unforgettable.

The more excited I got, when I set off for Brussels on Thursday, around half past seven in the morning. Everything went surprisingly smoothly. No delays, no breakdowns. Unfortunately, others were not so lucky. Malene from Denmark, for example, was stuck on the plane because she had to wait for her wheelchair. So she missed her scheduled train. Lucky for me, I met her in Brussels and we were able to continue our journey together.
In Bruges, Stephanie and Laurence were finally waiting for us at the train station to take us by car to the hostel. I cannot describe the feeling that came over me when I finally saw all the familiar and dear faces again. One by one, all 38 participants from nine different countries arrived safe and sound. And immediately you could feel the exuberant mood and good humour. 

After the first dinner together, specialties from the different countries, which some of us had brought with us, were distributed. Whether it was sweets from Norway, pretzels and sauerkraut from Germany, stroopwafels from the Netherlands or gin from England, everything was tasted. It was a wonderful evening with intense conversations. 

A sporty day one
Friday morning started sportily. Darts has become a kind of national sport in Belgium. Some professional players of the Belgian G-Darts national team came over to show us some of their tricks. Everyone was allowed to try their hand and even one or two "serious games" were played. Around noon, we set off together through the both beautiful and extremely bumpy streets of Bruges. The cobblestones were one of the most difficult challenges for each of us. But the beautiful half-timbered houses, the small canals and alleys provided a truly dreamlike backdrop.

Our destination was the Queen Astrid Park in the heart of the city. Here we could really enjoy the beautiful warm weather. With almost 30 wheelchairs and in yellow – it was Wishbone Day after all – we were a funny and probably very bizarre sight.

Charlotte from the UK offered a small “creative writing” workshop for those who were interested. Others went off to explore the city or enjoy the sun in a small café on the large market square. 

In the evening, another highlight awaited us: an exclusive beer tasting with traditional Belgian beers. In addition to fundamentally different tastes, there was interesting background information on the tradition and history of the Belgian art of brewing.

There was also a lot of laughter and sharing that evening. A few brave ones even tried the karaoke machine. The evenings together are the highlights of each day. Sitting together, talking and laughing, you sometimes get the feeling that time is standing still.

Puzzles and party
The next morning, on the other hand, began with sleepy faces. But anyone who has ever been to an OI meeting before knows: that's part of it! And luckily we had planned Saturday morning for some free time.

So everyone could start the day in peace. Some went for brunch, others preferred to recharge their batteries with a power nap or relax in the warm sun.
For Stephanie and me there was no time for sleeping. We had to prepare everything for the city game in the afternoon. Digitally, we went on a journey back in time to 18th century Bruges. Navigated by an app, the participants could solve puzzles in small groups and get to know special places in the city. Unfortunately, we had underestimated the cobblestones, the energy of our friends and the difficulty of some of the puzzles. In the end, only one group endured until about a third of the tasks were solved. The others quickly took a break in the restaurant or made themselves comfortable by the canal.

Yellow t-shirts everywhere!
Julian and I were also no longer mentally able to think logically, but we still wanted to explore a few beautiful sides of Bruges. Strangely enough, we kept running into tall and short people in yellow T-shirts, who looked at us with great interest every time. The Belgian OI Society was also in town, because we had planned a dinner together in the evening. The chance encounters quickly developed into a fun game, similar to Pokémon go, but with OI people.

The dinner exceeded all our expectations! At “LIO’s” we were joyfully welcomed by the Belgian OI Society. On the banks of the canal, there was a small welcome with various hors d’oeuvres and later a delicious three-course meal. At the party that followed, we danced and recruited new participants for the next OIFE Youth Event.

The way back to the hostel was adventurous. We were tipsy and wheeling in the dark over cobblestones (challenge accepted!). But this challenge was mastered without incidents and of course the party continued at the hostel!

Pain of farewell
The pain of farewell was diminished a little by the good mood, or perhaps just pushed aside, until finally everyone fell tired into their beds. Some of us had to start our journey home at 6.45 a.m. on Sunday morning. So what was the point of going to bed, we thought ... How could we have known that some of us would arrive home up to ten hours late. Nevertheless, I still think back to those four wonderful days with joy and longing.
My personal conclusion: Once again, it was absolutely worth it! I haven’t had such wonderful days in a long time and I am happy to have seen so many old friends and made new ones. At the same time, I am sad that the time has passed so quickly and, above all, that for some it was the last Youth Event. It will never be the same again.

Stephanie put it beautifully: “I have a fractured heart! I miss you all so much already.”

Important facts and figures:
- We were 38 people
- from 9 different countries,
- had 100% fun,
- were 85% sober
- and had 0 fractures

AND THAT DESPITE COBBLESTONES!!!

OIFE 2ND YOUTH EVENT 2022

Did you know that there will be 2 OIFE Youth Events in 2022? The 2nd one will take place in Warsaw, Poland from September 23-26.

Make sure to save the date. Registration will open soon.

Questions can be asked at oife.poland2022@gmail.com
Tell us about your experience with OI-work and what you are working on!
I joined OI-work in 2012 during my Master studies in The University of Tartu, Estonia. Afterwards I continued OI research in genetics, looking at genotype-phenotype correlations and clinical variability in OI families as a PhD student in the University of Tartu. Currently I am working as a post-doc researcher in Amsterdam UMC developing various therapeutic approaches for OI (gene therapy and pharmacological therapy).

Tell us about the project "Reproductive options"
During my time in The University of Tartu we had a very successful collaboration with Prof. Andres Salumets and Competence Centre of Health Technologies, which specialize on reproductive medicine and biology. His research group are pioneers in Estonia of preimplantation genetic testing (PGT) of various disorders including OI. Additionally, they were interested in developing of non-invasive prenatal testing (NIPT) for OI and other monogenic disorders. This is how the collaboration between an Estonian OI research team and the reproductive medical biologists started.

As we were looking closer at the topic of OI & reproduction it became clear, that literature regarding the latest reproductive techniques applicable for OI and associated ethical concerns was very limited. Both professionals and OI people are missing important information, which should help them to advice and make one of the most important decisions in life - family planning. The work was funded by three different grants to the University of Tartu, Estonian University of Life Sciences and Competence Centre on Health Technologies

What is the review about?
The review is an example of a multidisciplinary team effort, which appeared as a result of collaboration between experts in reproductive medicine, OI research and ethics. Our work systematized novel insights into OI genetics, reproductive options, family planning, decision-making and ethical issues. We aimed to fill the existing gaps in literature on OI & reproduction with our narrative review and bring the latest updates in advances of reproductive approaches for families with risk of OI to health care professionals, researchers and the OI-patient community in order to restore reproductive confidence of people with OI.

What was the purpose behind it?
As a rare hereditary disorder, OI is associated with numerous reproductive challenges, underlined with variable choice of reproductive options, decision-making and ethical concerns. During my interviews for the genotype-phenotype correlation study of OI families, I noticed that family planning, lack of clarity and confidence and sometimes poor experiences regarding this question is a burden. For the Quality of Life 4 OI conference in 2019, I was asked to give a talk about expectations and needs of OI people in the fields of basic research. Interviews with OI-people about important topics in research showed that the importance of family planning guidance was a big concern. OI-people need something more than “50% chance to pass on the mutation”, which is the same as flipping a coin. As a person with OI myself, I also spent quite some time exploring this topic and thinking about OI and reproduction.
What were your most interesting findings?
There is no one universal best reproductive option for all OI families. In the center of the decision should be the final outcome, which the person with OI set themselves, based on their desire, circumstances and abilities. Afterwards, different reproductive strategies should be carefully evaluated and a best approach for the case should be chosen together with professionals.

We have created a diagram of decision tree for families with (risk of) OI to give an overview of all options available for OI family planning. In this way we wanted to support OI families in decision-making. The diagram concludes all information and various scenarios in family planning. Based on type of OI and inheritance pattern, it leads you to chances of disease transmission to offspring, and shows approaches and techniques of modern reproductive medicine which can be used to ensure birth of kids not affected with OI.

What are the reproductive approaches suitable for prospective parents faced with a risk of OI?
First of all, ethical and supportive attitude of clinicians is crucially important for OI patients, as family planning is extremely sensitive. We should spend more attention on sharing information about reproductive options with patients. In collaboration with patient organizations, we should improve patient education so that autonomous decisions of people with OI would be fully informed.

Early family planning, pre-pregnancy OI genetic testing or pre-carrier screening will increase availability of various options of fertilization and prenatal testing. For fertilization a few options are available: natural conception, in vitro fertilization (IVF) with donor cells or embryo, or preimplantation genetic testing (PGT-M). All methods have advantages and risks, thus each method should be evaluated, with a special attention given to the wishes, interests, health needs and opportunities of the OI person.

The field of rapidly evolving non-invasive prenatal testing (NIPT) techniques should be constantly followed. Although detection of OI in unborn babies where the mother is affected with OI, is still studied (soon I expect it to be available), detection of
de novo OI and OI in unborn babies with an affected father are ready to use options in many locations. Non-invasive testing options such as NIPT and ultrasound should prevail for prenatal diagnosis. Prospective parents’ decisions regarding invasive prenatal testing methods such as CVS, amniocentesis and cordocentesis, which carry risks for the pregnancy, need to be made autonomously, but with support.

What are the ethical aspects?
Reproduction for people with genetic disorders is a very sensitive topic. The biggest concern is autonomous decision making and its consequences. Very important OI-specific ethical issues are a correct prediction of OI severity (clinical type) and lethality based on the genetic mutation, which is currently impossible due to lack of genotype-phenotype correlations, and additional yet to be studied factors which cause clinical OI severity. There is definitely insufficient research on fertility in OI patients, soft tissues in OI pregnancy, lack of ethical studies related to anxiety and stigmatization around reproductive decisions in OI families.

Did you have patient involvement in the project?
We involved a representative from OIFE in reading and reviewing the article text, because we wanted it to be clear to the community. We also revised carefully a decision tree diagram with support from the patient community. Although some parts of the review are more complicated for general reader, we hope that the OI-community can also directly benefit and use this work. We would like to thank Ute Wallentin for her comments and suggestions, which improved our work.
There is a lot of confusion around OI and inheritance. Do you have a simple and pedagogic way to describe the different types/classifications of OI in not too many words?

First of all, there are two types of classifications. The clinical types (OI types 1-5) and genetic types (OI types I-XX+). These are two different classifications, and none of them are perfect. If we can simplify the model, clinical classification – is like a rainbow. We have 5 types, clearly different (like colors in rainbow we can clearly see), however there are also many border-line types. It’s the same with rainbow, it is sometimes hard to say were exactly one color changes to other. See the illustration on the previous page for more details!

Genetic classification is like an iceberg. The tiny tip of it are only two genes, both for collagen 1 (COL1A1, COL1A2). And ~85% of OI people are on this tip. As a rule it is dominant OI, in other words – one mutation is enough for OI. For those OI people from the tip of an “iceberg” it is 50% chance of passing OI to offspring. The bottom huge part of an “iceberg” are various other recessive genes (now we know ~20 of these OI genes). In the bottom part there are only ~15% of patients. If you are among the “iceberg bottom”, it is usually recessive OI. For recessive OI two mutations altering the same gene are needed. Risk of OI transmission is 25% if both parents are carriers (have one copy of the mutation each). There are also some exceptions, like type 5 (clinical). This is always due to the IFITM5 gene, dominant mutation, so 50% chance.

Any messages for the readers of OIFE Magazine?
The most important is what you want. Based on your own desire, go for professional support and together with them build a strategy. Reach for information, educate yourself, understand the risks, learn what is acceptable for you and what is not. If some of the services are not available in your country, reach expertise centers in neighboring regions. Here are top 5 take home messages for the OI-community:

- ~85-90% of OI patients have a 50% risk of OI transmission to the offspring (dominant OI).
- More than half of OI-cases appear without previous OI-history in the family (sporadic/de novo).
- There are people who do not have OI, but have a high risk of OI transmission to offspring (carriers of recessive OI mutations, parental gonadal mosaicism).
- If a parent has dominant OI, a prenatal genetic testing (PGT-M) can help to have a non-OI baby.
- It is possible to perform testing (NIPT) of OI in an unborn via a pregnant mother’s blood sample.
Fetal diagnosis of OI during pregnancy

Interview with Shannon Bonner, graduate student of genetic counseling at Baylor College of Medicine, Houston, Texas, USA

Who are you & what is your relationship to OI?
My name is Shannon Bonner and I am currently a graduate student studying genetic counseling at Baylor College of Medicine which is located in Houston, Texas. In the coming weeks, I will be graduating with my master’s degree in genetic counseling, and I will be starting my career as a prenatal genetic counselor at Texas Children’s Fetal Center. I first became involved with the OI community when I began exploring the topic of my master’s thesis research. Considering my interest in prenatal genetics, I was excited when the opportunity arose for me to investigate the experiences of parents who receive a fetal diagnosis of OI during pregnancy.

Who was behind the project? How was it financed?
I was first introduced to this research project by Chaya Murali, MD, a pediatric medical geneticist who sees patient’s in both the general genetics and skeletal dysplasia clinics at Texas Children’s Hospital. Dr. Murali and I developed this project with the help of several researchers at Baylor College of Medicine in addition to Michelle Fynan, PhD, who is an active member of the OIF. We could not be more thankful for the help we received from both the OI Foundation and the OIFE who aided in recruiting participants for this study. This research was supported in part by a research grant from the Genetic Resource Association of Texas.

What was your research project about and which methods were used?
My master’s thesis research is entitled, “L”. During our initial review of the literature, we identified an obvious gap in research surrounding how to best counsel and support parents when they receive a diagnosis of OI during pregnancy. Guidelines exist that aid medical providers in diagnosing and managing OI in the prenatal setting; however, there is no literature that addresses the emotional aspect of receiving this diagnosis prenatally.

To address this gap, we conducted a mixed-methods study which utilized a brief online survey and telephone interviews to gain insight into parents’ experiences with prenatal genetic counseling and how satisfied they are with their overall experience. We recruited individuals for this study who received a prenatal diagnosis of OI within the last five years and were subsequently counseled by a genetic counselor, medical geneticist, OB/GYN, or maternal fetal medicine specialist.

What were your most interesting findings?
Ultimately, we found that parents receiving a prenatal diagnosis of OI are generally less satisfied with their genetic counseling experience when compared to other populations receiving prenatal care, such as those with abnormal prenatal genetic screening or individuals at increased risk for chromosome conditions in a pregnancy. When parents receive an OI diagnosis during pregnancy, yes, the medical management and informational aspects of the diagnosis are important. However, the parents from our study expressed a desire for counselors who recognize the challenge of navigating the uncertainty of this diagnosis and focus more on fostering hope and enhancing parental coping.
Through the interview portion of our study, we identified several factors that influence parental satisfaction with counseling, and we hope to discuss these findings in more detail through a future research publication. Briefly, we observed that parents valued counselors who provided comprehensive, tailored counseling which included discussions about all possible pregnancy outcomes, despite provider predictions of lethality. Parents also desired immediate referrals to OI community resources, such as the OIF and OIFE. Parents with a personal or family history of OI often described experiences where their counselors held preconceived assumptions about their knowledge of OI. Thus, we encourage counselors and other clinicians to provide comprehensive counseling to these individuals, acknowledge their lived experiences, and avoid making assumptions about their knowledge of caring for a newborn with OI.

Any messages for the readers of OIFE Magazine?
I’m grateful for the OIFE for distributing information about our study during recruitment and providing me this opportunity to discuss my research. I am also incredibly appreciative of all our research participants for sharing their stories. This project has played a pivotal role in my journey to becoming a genetic counselor and the lessons I’ve learned from the OI community will undoubtedly have a lasting impact on my practice.

“The role of inheritance”
Knowledge in family planning among young adults with OI

Interview with Leanne Baird, graduate of the University of Cincinnati Genetic Counseling Graduate Program

Who are you & what is your relationship to OI?
My name is Leanne Baird, and I am a recent graduate of the University of Cincinnati Genetic Counseling Graduate Program in the US. I have a passion for educating patients about genetic risks and conditions, as well as helping patients navigate difficult decisions in relation to said genetic risks. Due to these passions and OI being a genetic condition, selecting this project as my thesis was an easy choice.

Who was behind your research project?
Dr. Kara Ayers, PhD, my research advisor on this project who has OI type V herself, mentioned many misconceptions amongst individuals who have OI. This sparked a curiosity in us to determine OI inheritance knowledge levels within the young adult OI community, as we wanted to see if knowledge would impact future reproductive choices. Jodie Johnson, MS, CGC is a genetic counselor who has worked closely with many patients and families affected by OI over the years. Jodie was able to provide personal clinical experience to this project.
The final member of our team is Nichole Nidey, PhD, who provided her statistical expertise to pull interesting results from this our data.

What is this research project about?

Objectives:
1. Evaluate OI inheritance knowledge levels in young adults with a self-reported autosomal dominant or unknown OI type
2. Determine from who and when OI inheritance information is learned
3. Investigate the role of OI inheritance in future reproductive decisions

Target Group:
We included those who were 18 - 25 years old, with a self-reported autosomal dominant (types I-V) or unknown OI type. All participants had to have the ability to read and understand written English, as study materials were only available in English.

Methods:
An author developed questionnaire was distributed to participants through the OI Foundation and OIFE newsletters, research blasts, and social media pages. The questionnaire was composed of 4 sections; part A collected demographics and evaluated for inclusion criteria, part B calculated total OI inheritance knowledge scores, part C determined OI inheritance information sources and the age participants 1st remember learning this information, and finally, part D addressed desire to have children, the influence and impact of differing factors on reproductive decision-making, and interest in reproductive options.

What are the most interesting findings & how may they impact care in the OI community?
Young adults with an autosomal dominant OI type have a good grasp on basic inheritance information, but knowledge gaps have been exposed that may require additional education, and there seems to be difficulty in applying basic inheritance information to more complex real-life situations. The topics that show a need for increased education in this community include:
- OI does not skip generations
- The gene affected influences the OI type an individual has
- Chance of OI in a future child when both parents have OI
- Chance of a genetic condition in a future child when one parent has OI and the other has a different autosomal dominant genetic condition

Parents are the primary, and a reliable information source of OI inheritance information. Most participants report learning about the inheritance of OI under 10 years old, but the age of first learning this information does not impact knowledge level.

About half of our participants want to have a biological child in the future, with the inheritance of OI and personal physical experience with OI being the largest deterring factors. Participants are very interested in prenatal diagnosis and IVF with preimplantation genetic testing of monogenic disorders (IVF+PGT-M) but are not at all interested in using donor eggs/sperm. Interestingly, as OI inheritance knowledge level increases, interest in prenatal diagnosis decreases. Participants who reported inheritance as the most influential factor over reproductive decisions trend toward having less interest in prenatal diagnosis, more interest in donor use, and has no effect on interest in IVF+PGT-M. This study shows potential concerns and considerations that could arise when caring for young adults with OI who are making difficult reproductive decisions.
Pregnancy in women with OI & osteoporosis

By Claudia Finis, mother with OI and Prof. Dr. Ralf Oheim, Chair of the National Bone Board at Universitäts-Klinikum Hamburg-Eppendorf, Germany

Claudia: I can still remember the time when I felt like a big whale on four wheels meandering through the world. A vague idea about the existence of my knees and permanent heartburn accompanied me 24/7. I had no problems with my bones or heart and could breathe properly. The births, however, were not without complications. But was that because of OI? Or simply because anything that happened to me can happen to anyone?

Every now and then the German OI-organization receive questions about pregnancy in women with OI. There aren’t a lot of studies on this subject. Some researchers, including Dr. Krakow, conducted a study in the USA. I took a closer look at the study “Pregnancy in women with Osteogenesis Imperfecta: pregnancy characteristics, maternal and neonatal outcomes”. Dr. Krakow also gave an interesting (online) lecture at the OIF conference.

Dr. Krakow’s study
First of all, Krakow recommended that women who want to have children have their cardiovascular system and lung function checked. If necessary, operations should be performed before pregnancy. The heart and lungs face special challenges during pregnancy. The best possible starting position should be ensured.

For the study, retrospective data were collected from 132 women over 170 pregnancies. 74.4% of the women rated themselves as mildly affected, 24.0% as moderately, and 1.6% as severely affected. 48.9% said they had no mobility restrictions, 48% said they had some restrictions, and 4.1% were wheelchair users.

Of the women who classified themselves as mildly affected, 43.4% had a vaginal birth, while only 8.11% of the women with moderate or severe OI had a vaginal birth. Surprisingly, 41.3% of babies with an OI were delivered vaginally, while only 25.64% babies without an OI where delivered the same way.

Can women with OI have successful pregnancies?
Women with OI have successful pregnancies. The study concluded however, with pointing out an increased risk of complications including bleeding, fractures, diabetes and increased neonatal morbidity. Women with OI have a fracture rate of 10 to 12% during pregnancy and after delivery, according to the study. The fractures occurred in the third trimester and 2 months after birth. The spine was most affected, especially in the period after the birth. Medical staff should be informed of the increased likelihood of bleeding. Dr. Krakow recommends starting the appropriate measures earlier than usual in order to avoid risks.

How is the situation for women with OI in Germany? Interview with dr. Ralf Oheim
There is no comparable study in Germany. However, Prof. Dr. Ralf Oheim drew my attention to a study that initially looked at pregnancy osteoporosis. In the course of collecting the data, the researchers, discovered that some of the women affected had mutations in the COL1A1 and COL1A2 genes.

Commonly these are also mentioned when it comes to OI. But the women showed no clinical signs of OI. The mutations were not found until the women participated in the study. I think this is a very exciting discovery. So I decided to interviewed Dr. Oheim about bone health in pregnancy and about the study.
Claudia: In the third trimester the mineralization of the bones of the fetal bones begins. During this period, the baby needs more calcium, which has to be made available by the mother and if necessary, can also be taken from the mother’s bones. It is therefore not surprising that in both of the studies mentioned here, the fractures did not appear until this phase of pregnancy?

Ralph: Especially in the 3rd trimester of pregnancy and during breastfeeding there is sometimes greatly increased bone metabolism. The calcium absorbed through food can still be stored in your own bones, but when it’s critical, the child always has priority. This can create a deficit for the maternal bones. The bone-degrading osteoclasts are always faster than the bone-building osteoblasts.

Claudia: Is there a minimum value which prevents the calcium in the maternal bones from falling below?

Ralph: No. The mother’s body does everything in its power to keep the calcium level in the blood within the normal range, as this is vital for the mother herself. If the calcium supply through food is not ensured, bones are broken down in order to release the required calcium. The calcium level in the blood therefore unfortunately only provides very limited information on whether the calcium supply is sufficient. A twin pregnancy is also a particular challenge for expectant mothers in terms of calcium supply.

Claudia: Does the maternal bone regenerate? If so, how long does it take?

Ralph: The study found that the bones recovered well within 6-12 months.

Claudia: Can the pregnant woman increase the calcium in the bones before the (planned) pregnancy? Or should the pregnant women also take calcium even if the value is normal and good?

Ralph: No, this does not have any positive effects. You cannot build up a calcium deposit in the run-up to pregnancy, as excess calcium is excreted. An oversupply of calcium should also be avoided during pregnancy and breastfeeding and the intake of calcium supplements in addition to a balanced diet is usually not necessary.

Claudia: In your article, women with pregnancy osteoporosis were given vitamin D. Would it make sense to take this prophylactically?

Ralph: Yes, if necessary, because a good vitamin D level (> 30µg / l) should be available during pregnancy. In the case of supplementation, a daily dose should be taken during pregnancy.

Claudia: Can medications against possible osteoporosis be taken during pregnancy and breastfeeding?

Ralph: Currently, none of the drugs are approved for use during pregnancy and breastfeeding in Germany. In the case of high-risk patients or fractures, it should be decided quickly whether breastfeeding is possible.

Claudia: In the literature there are repeated reports of osteoporosis or fractures of the hip during pregnancy. What precautions should I take due to the risk of hip osteoporosis?

Ralph: Cancellous bone is preferred in bone resorption because of its large surface area, problems are often caused by the hips and vertebrae. But one should definitely keep moving. Immobility promotes bone loss.
Complaints should, however, be taken seriously and not just viewed as normal pregnancy symptoms! There should be an individual review. For the benefit of the child, diagnosis is only possible to a limited extent, but an MRI can be performed in an emergency. After the delivery, the corresponding diagnostics are possible without restrictions and should be carried out generously.

Claudia: In the studies, fractures did not occur until the last trimester of pregnancy. How is a fracture (e.g. femoral neck) treated then? Are operations possible? Will it lead to an early end of the pregnancy?

Ralph: Conservative treatment with rest and relief is preferable and often possible, since the fractures are often incomplete and not displaced. In the third trimester, the placental barrier is well developed and, if it is unavoidable, an operation can be carried out during pregnancy. If the calculated due date is imminent, the birth can also be brought forward. The choice of these alternatives must be considered individually.

Claudia: Contrary to the reports from Dr. Krakow’s study, all mothers with OI I know gave birth by caesarean section. Is it possible to have a natural birth with OI? Does this depend on the type of OI?

Ralph: I know Type I mothers who gave birth naturally. With types III and IV, on the other hand, the anatomical conditions on the maternal side often lead to the decision to have a caesarean section.

Claudia: And now the baby is born. Breastfeeding is considered to be the best form of nutrition for infants. If mothers now decide to do this, they are threatened with major loss of calcium from their bones. What can be recommended to the mothers in this case?

Ralph: If the pregnancy and childbirth proceeds without complications, nothing speaks against breastfeeding for the time being. In high-risk patients, the bone density and bone metabolism of the new mothers can be checked after about two to six weeks and, if possible, compared with values before the pregnancy. On the basis of these values, the physician and the mother will together decide whether or not it is possible to continue breastfeeding. A good vitamin D level (> 30µg / l) should also be ensured and, if necessary, it should also be given daily.

Ralph continues: In the case of fractures, breastfeeding usually must end quickly, or breastfeeding should not take place in the first place, in order for the bone to heal and recover. Medication is only necessary in severe cases to avoid further aggravation and to enable early mobilization.

Claudia (addressing the readers): I hope you are not scared off now, but well informed for your decision. Every pregnancy is a risk. If you want to face them, you are welcome to read about other aspects of being a parent with OI in my article „Having OI and being a parent“ in OIFE Magazine 3-2021. Even if everything did not go smoothly during the births, I do not regret my decisions. I know that I am not alone with this opinion. In the end, every woman and every couple has to make this decision for themselves.

Claudia and her husband
USA Case Discussion project for orthopaedic OI-surgeons

Interview with Dr. Jeanne Franzone & Dr. Richard Kruse,
Nemours Alfred I. duPont Hospital for Children, Wilmington, Delaware, USA

Soon after the initiation of the COVID pandemic, in June of 2020, Dr. Jeanne Franzone (Nemours Alfred I. duPont Hospital for Children, Wilmington, DE), Dr. Maegen Wallace (Children’s Hospital and Medical Center Omaha, NE) and Dr. Jill Flanagan (Children’s Healthcare of Atlanta, Atlanta, GA) initiated an OI Surgeon Quarterly Case Review: A Virtual Forum.

The forum was developed to facilitate a virtual discussion among surgeons with an interest in OI surgery, to meet to discuss challenges related to OI surgeries. Drs. Franzone, Wallace and Flanagan all specialize in the orthopaedic care for patients with OI as part of multidisciplinary OI programs and sought to gather surgeons with an interest in and a passion for improving the surgical care for patients with OI.

What kind of case discussions do you have?
We meet on a quarterly basis by Zoom and we discuss OI-cases submitted by the participating surgeons – these may be completed surgeries or upcoming surgeries that demonstrate a learning point. Surgeons may submit cases to seek advice. We discuss both broad topics as well as the detailed aspects of OI surgery. We also place importance on follow-up discussion of prior cases so that we all continue to learn.

Who can join?
We aim for this to be an inclusive environment, and given time constraints, each surgeon may not be able to attend every meeting. We currently include 33 surgeons from 22 institutions in the US. We emphasize that it is a respected environment in which challenges and complications may be discussed.

How do you deal with anonymization and privacy?
It is very important for patient privacy to be respected. We ask for the cases to be deidentified of patient names and identifying information. We emphasize that it be a respected environment.

Have there been any challenges?
Absolutely! In addition to the usual technical challenges, a challenge in gathering a group of busy orthopaedic surgeons is finding the time and finding a time of day that works across different time zones. For this North American based group we currently meet in the evening hours to try to steer clear of morning conferences, patient clinics and planned surgeries.

What are the lessons learned so far?
We have had many interesting discussions regarding surgical indications, techniques and implant considerations. As noted above, the follow-up provided regarding the cases discussed greatly enhances the learning opportunities.

Would you recommend other countries or regions to establish similar projects?
Yes. We have found this type of virtual discussions to be a valuable way to bring together surgeons from different institutions to share ideas and learn from case-based discussions. There is benefit to collegial discussion among surgeons specializing in OI, to incorporate care within their specific healthcare system.

Do you ever publish some of these cases as case studies?
The US-based OI Quarterly Case Review has been a clinical discussion, more case-specific.
How could we make sure that knowledge benefits orthopedic surgeons in less developed countries?

By sharing this information here, we hope to encourage similar discussions in different healthcare systems around the world. We (Dr. Kruse and Dr. Franzone) have also initiated an international forum to include different healthcare systems and resources. We also encourage similar forums among other specialties and care disciplines.

Please tell us more about the international forum!

We have organized an International OI Surgeons Work Group. This is an international gathering of specialized OI surgeons including surgeons from 7 countries and 3 continents. The initial meeting took place in May 2022. It was an exciting gathering, very dynamic and interactive and set the stage nicely for international collaboration of OI surgeons regarding challenging topics such as acetabular protrusion. Although also a case-based discussion, the aim for this group is to discuss surgical care in a way that spans different healthcare systems and available resources. This international perspective will be used as a springboard to facilitate international research collaboration and knowledge dissemination. People who have questions about this forum can contact office@oife.org

Activist with OI: Umi Asaka

Umi, who is originally from Japan, moved to New Zealand after the nuclear disaster in Fukushima back in 2011. Today, she is living in an accessible house on South Island together with three friends. Umi talks about her life and what it means to live in New Zealand as an immigrant with a disability.

My name is Umi Asaka (25). I am originally from Japan, and I moved to New Zealand almost 10 years ago after the nuclear power station’s accident in Northern Japan. I am a daughter of a strong disability activist, Yuho Asaka. I inherited OI from her. I think my relationship with OI has been quite unique because of this. I have broken mainly my left femur throughout my childhood, but I have never had surgeries for them, and I have not broken them since puberty. I always had a role model of how you can navigate this world and look after our bodies. It makes me think that if have a child, I would like to have a child who shares our condition, so that we can share our experiences with them. But of course, they can be born with or without OI or anything and that is great.

Can you describe a little where and how you live?

Currently I live in Dunedin which is South of South Island in New Zealand. I live with three other friends and out of four people, three of us are wheelchair users. Since it is my friend’s house it has been possible to modify the house to be accessible for us. All of my housemates can drive a car, and I am slowly learning to drive. One of my housemates has kindly let me use her modified car to practice driving in. I usually commute to my work by bus, and it is relatively accessible. By relatively I mean sometimes the ramp on the bus is very steep and I don’t feel safe. But I have been able to get on and off the bus. Major places are accessible, but of course there are cafes, restaurants and other venues that have odd steps to get in without any other access options.
How is the situation of people with a disability in New Zealand?
I think the situation in New Zealand is a mixed bag. You might have an image that New Zealand is a welfare country, and it is to some extent. The most positive thing is that big institutions have been closed. However, the health system and housing issue have been challenging for the whole community, and disabled people are still overrepresented in lower-income bracket. There is government funding for equipment, assistance and medication. However, the number of people eligible to access these are very cut-off and the waiting time is very long. There is actually no legislation for accessibility nor anti-discrimination. The government of New Zealand has announced an establishment of a Ministry of Disabled People (tentative name) and accessibility legislation within this year. The disability support system largely operates under western and coloniser’s perspective, so it often does not cater for the experiences of Māori (indigenous) disabled people. The public transport within a city is relatively accessible like I said, but there is no accessible transport between cities except for flying or driving yourself.

What do you do?
I studied social work at University of Otago, and I am working as a Junior Research Fellow at the Donald Beasley Institute (DBI). DBI is a national independent research institute for disability. One of our projects is Disabled Persons-Led Monitoring of UNCRPD (United Nations Convention on the Rights of Persons with Disabilities), where we interview disabled people across the country about their experiences relating to the specific topic we are monitoring at that time. Disabled people’s experiences are still quite invisible in the world, and our aim is to highlight the experiences so that much needed changes can happen. You can find more about the project here.

Are you active in an OI-organization?
I have a group chat with OI friends back home in Japan, but I have not been able to connect with OI community in New Zealand, and I would love to do so.

“I went to support the bus driver’s action towards fair pay as I use the bus every day. Solidarity between movements is important.”
Tell us about your activities as an activist!
I have been part of a few different activisms, from climate actions, peace building actions to disability activism. The most recent one I have been part of is #EndASHNow!. In Immigration New Zealand, there is a policy called Acceptable Standard of Health (ASH) requirement, which effectively rejects people with disability and health condition’s visa applications if they are not granted with medical waiver. I have been part of the movement to scrap this ableist policy, and we have been supporting individuals and families who are impacted by this policy. You can find more about our work through @endashnow on twitter or on the webpage. This policy is completely unacceptable, and at the same time, I think our action is revolutionary. The immigration policy had eugenic idea from the beginning, and we are challenging that underlining discrimination that has managed to stay in this society until now.

What can we do as individuals to change the world for the better?
I think what we can do is to live the way we want to live. Just like the feminist slogan “personal is political”. Our actions to thrive to live better lives are effectively paving the path for next generations of OI and other disabled people to live better lives, just like my mother have paved the path for me. In order for us to live well, we cannot avoid confronting ableism, and for me racism and sexism. It can get tiring at times, but you also meet lots of people who are fighting the same fight, and the energy you gain from having these people in your life is invaluable. So I do encourage young people with OI to get out and learn about how ableism all the other oppression impact our lives, and to find people who can journey and navigate the world with. I could not do what I do right now without the people in my life, and having that community and support network is so crucial.

Any messages to the readers of the OIFE Magazine?
Although I have not been able to be part of OI community for a while, I love being around OI people. I love our optimism, cheekiness and strength. If you want to connect with me, I would love to hear from you 😊 @asaka_ocean – twitter

“Being in nature is what I love, and New Zealand is a great place for that! Although many places are not so accessible, and I have friends who take me out into the wild.”
Who are you and what is your relationship with OI?
My name is Léa, I am 22 years old and I have OI. As a child and teenager, my family and I went to many OI events at the regional association in Germany. When I moved to the Netherlands for my studies, I got to know the Dutch OI association, which was a great experience. I find it very enriching to build an international network of people living with OI. It’s a great way to meet people from different countries and help each other.

Tell us about what you do!
I am studying Global Project and Change Management, a program that prepares me to become a project manager to tackle global challenges. Besides studying, I love cooking plant-based foods and inviting my friends over. I also enjoy reading books by Elif Shafak or Kübra Gümuşay. And holidays, but who doesn’t like holidays at the beach?

Right now I’m an intern at Rare Diseases International (RDI), which is part of my studies. Ingunn Westerheim sent me the internship posting from RDI online and encouraged me to apply. RDI was interested in my profile and they found it valuable that someone would do research for them. This is how I am currently writing my bachelor’s thesis at RDI. The purpose of my internship is to produce valuable research for RDI to build upon and continue their advocacy efforts. I started the internship in February 2022 and will finish in June 2022.

What are you doing there?
I study the commonalities and the differences between living with a rare disease and living with a disability. Therefore, I examine the challenges that people with a disability and persons living with a rare disease face and the impact this has on a person’s life. After analyzing the literature, I have begun to interview experts and patient experts in the field of disabilities and rare diseases. Through a survey, I would like to have rare disease experts prioritize and rate the overlapping and distinguishing elements. I will summarize my findings in a research report and an advisory report for RDI.

What are the three most important things we should know about the UN Resolution?
The Resolution "Addressing the Challenges of Persons Living with a Rare Disease and Their Families" is the first United Nations Resolution to address the specific challenges of persons living with a rare disease. Recognition is a first step in raising awareness and working toward change. I think this is something to be excited about and proud of as a rare disease community.

This achievement is the result of all the hard advocacy and political work of civil society, activists, and NGOs who have pushed for the recognition of the challenges faced by persons with rare diseases! Cheers to them!
The resolution is just the beginning, it now needs to be implemented by different countries and networks. It is up to them to develop their strategies and national work plans, etc. to improve the quality of life of persons living with a rare disease. And it is up to us to advocate for it.

OIFE had a campaign with the keyword EQUITY. How do you define equity for people with RD? For me, equity is not only an endpoint but also a means. People living with a rare disease (PLWRD) must be involved in decision-making to be fully represented and to develop solutions that truly serve PLWRD and their families. In addition, human rights must be respected. PLWRD should have the opportunity to receive a diagnosis and appropriate treatment. In addition, education, decent work, and a life free of poverty and hunger should be guaranteed. To ensure this, the appropriate resources to improve the quality of life must be provided.

How is the resolution connected to the CRPD or the UN development goals?
The UN 2030 Agenda with its 17 Sustainable Goals (SDG) calls for justice and equity in various areas. Discrimination starts early for children living with a rare disease, for many of them access to Quality Education (SDG 4) is denied or impeded. Families with a member living with a rare disease are at greater risk of impoverishment (SDG 1 No poverty) due to higher expenses to care and fewer earnings because one parent often has to reduce working hours or stop working to provide care.
Women are disproportionately discriminated against and excluded from society (SDG 5 Gender Equality) either as patients themselves, in family planning or as the main caregiver of PLWRD and her family. SDG 8 Decent work is impeded for PLWRD as they experience difficulties in the process of independent living to finding, keeping, or returning to work after hospitalization.

The UN resolution, therefore, refers not only to the Conventions on the Rights of Persons with Disabilities (CRPD), but also to the Universal Declaration of Human Rights, the Convention on the Rights of the Child, the International Covenant on Economic, Social and Cultural Rights, and the Convention on the Elimination of All Forms of Discrimination against Women.

**What kind of effect do you hope the resolution will have?**
I hope that the resolution will open the eyes of policymakers, politicians, and those in power. Health is a human right and should be accessible to everyone, including persons living with a rare disease. The rarity of the disease should not be a criterion for discrimination and a lower quality of life. Even though we still have a long way to go, I am sure that a better and more equitable world is possible!

**What are your dreams for a job/career?**
I don't know what the future holds. What I do know is that I want to pursue a career that has a positive impact on the world and our society. Although there have been improvements in women's rights, disability rights, and PLWRD quality of life over the past few decades, we still have a long way to go before we achieve full equity for all people. I would love to work in an organization that addresses systemic injustice and strives for intersectional justice.

**What should OIFE focus on in our advocacy work?**
Well, I'm not an expert on advocacy. But I think inclusion and diversity are very important for advocacy. People with OI face different challenges depending on where they live, when they received their diagnosis, what impairments they have, and socioeconomic differences, among many other things. These differences must be embraced and considered when advocating for people with OI. OIFE's responsibility is to listen to these different experiences, to include people of all genders and backgrounds, and to be open to diversity and a wide range of knowledge and experience. Only by being inclusive can we reach a diverse network of people.

**Any messages for the readers of the OIFE magazine?**
Follow your heart! Be kind!
News in brief

**OIF CONFERENCE 2022**
Are you planning on joining the annual conference of the Osteogenesis Imperfecta Foundation which will take place online this year as well? Program starts 16.30 Central European time.

**REHABILITATION FOR OI**
After several years of advocacy from the Norwegian OI-group Norsk Forening for Osteogenesis Imperfecta (NFOI) we finally succeeded in having an annual rehabilitation course for adults with OI in Norway at Rede Kors Haugland Rehabiliteringssenter AS. Our GPs prescribes 3 weeks specialized rehabilitation, and you get paid sick leave or your normal disability pension while you are there.

**GOOD LUCK REBECCA!**
We wish Rebecca from OIFE best of luck with her new position as Board member of EURORDIS - European Rare Diseases Organisation!

**NEW OI DIAGNOSIS TOOLKIT**
The Osteogenesis Imperfecta Foundation has together with dr. Michael Bober created a toolkit for parents and medical professionals treating a new OI diagnosis. This video is the first in a series of videos that will accompany the OIF’s Navigating a New Diagnosis Tool Kit. The New Diagnosis toolkit and video resources are available on the OI Foundation’s website: [www.oif.org/newdiagnosis](http://www.oif.org/newdiagnosis).
LATIN AMERICAN OI EMERGENCY CARDS
Several OI-organizations in Latin-America have collaborated with the support network Conectiva (Hereditary Connective Tissue Disorders) to develop an emergency card for OI. The main purpose of the emergency card is that through the QR code or the web link it incorporates, health professionals can access an emergency protocol of OI created by medical specialists. The cards are free of charge and can be downloaded for free at the website:  https://conectiva.lat/emergencias/

HOUSING FOR UKRAINE REFUGEES
The rare disease umbrella EURORDIS - European Rare Diseases Organisation has teamed up with Airbnb.org (supported by Airbnb) to provide free and safe housing abroad for Ukrainians with rare diseases who are fleeing the country because of the Russian invasion. If you or a member of your immediate family are in need of a safe place to stay outside of Ukraine, please visit: http://l.eurordis.org/jt15

SURGICAL EQUIPMENT
Two of the companies producing surgical equipment for children with OI have decided to merge. We hope to continue the good collaboration with Pega Medical and OrthoPediatrics.

REPORT ON TRANSITION FROM PEDIATRIC TO ADULT CARE IN THE UK
Young people living with rare conditions in the UK share their experience of transitioning from paediatric to adult care in this new report created by Costello Medical in collaboration with Cambridge Rare Disease Network & Beacon for Rare Diseases.

We believe it can also be relevant for other countries. Download & share https://lnkd.in/e4zc5WZ6

GLOSSARY FOR MEDICAL TERMS
Do you think medical language is complicated and hard to understand? Then you are not alone. The European Medicines Agency (EMA) has developed a substantial glossary to explain medical terms connected to medicines and medical treatments with plain language descriptions. You can download the glossary for free here: https://bit.ly/3nsazwQ
CONGRATULATIONS MCKENZIE!
June 13th — one day before her 26th birthday — McKenzie Coan from the US captured her third consecutive world championship in the 400-meter freestyle at the Para Swimming World Championships in Madeira, Portugal. McKenzie already owns gold medals in the event from the Paralympic Games in Rio de Janeiro in 2016 and Tokyo in 2021.

COURSE FOR YOUNG PATIENTS
Are you between 12 and 18 and interested in health, biomedical research, healthcare and children’s rights or a patients with a rare disease interested in making research more suitable for children? Then Check out the European Joint Programme’s Training for Paediatric Patients at the 2022 iCan Summit from July 12-15 at the University of Lyon in France. Accommodation and travel costs are fully covered. The deadline to apply is July 1st. cutt.ly/iKm5BVm

Get in touch!

The Osteogenesis Imperfecta Variant Database (OIVD)
The Oi Variant Database has been serving the OI community for decades by providing a reliable source to consult about the pathogenicity of genetic variants. OIVD owes its creation in 1984 to Prof Raymond Dalgleish who dedicatedly ensured its continuation until his retirement in 2021. Thanks to the critical involvement of OIFE, the curation of the database has recently passed to Amsterdam UMC and is now hosted by the Leiden Open Variation Database (LOVD). OIVD currently contains approximately 3250 unique OI variants in 6290 patients, with complete variant listings until 2018.

OIVD is acknowledged worldwide as a powerful tool assisting OI professionals with cases of genetic diagnosis, prediction of disease progression, genotype-phenotype correlations, and translational and clinical research. Considering its enormous significance in the OI field, the curation team of the Human Genetics department in Amsterdam UMC (Dimitra Micha, Gerard Pals, Sonna Stolk, Lidia Zhytnik, Marelise Eekhoff and Taco van Welzenis (OIFE)) has committed to its maintenance in the future. Raymond Dalgleish remains in an advisory role. Currently, OIVD is being updated with missing OI variants from 2019 to the present, which will also include 930 variants from the database of Amsterdam UMC.

OIVD is the only database dedicated to OI, in which genetic variants are evaluated by experts in OI molecular genetics. As such, it provides reliable information about their molecular significance in combination with the clinical presentation. We want to strongly encourage OI professionals to continue submitting their identified variants in the database (https://lovd.nl/OI-genes) to promote consolidation and harmonization in OI variant interpretation. Variant submission can be easily done by registering in OIVD as submitter. We warmly thank the support we have received for this initiative from the Osteogenesis Imperfecta Society Australia.

Dimitra Micha
Results from the OIF’s survey on Covid19 and OI

To have more knowledge on the health issues of people with OI, it’s important that we contribute with our experiences through surveys and other ways of collecting data. 577 community members completed the American OI Foundation’s third survey about Covid-19. Some of the main findings are shown below. For more information – check out oif.org
OIFE Investigator Meeting 2022

1ST VIRTUAL OIFE INVESTIGATOR MEETING

18TH NOVEMBER 2022
2 PM - 7 PM CET / 8AM - 1 PM EST

OUR AIM
Our aim is to present an overview of current OI-research (basic and clinical) in Europe and beyond. Presenters will include both experienced OI-researchers and new investigators. The event will be a supplement and a follow-up to the 14th International Conference on OI in Sheffield in September.

TARGET GROUP
The target group is primarily researchers and clinicians working with OI, but anyone interested in OI-research can join.

Further information & updates
For further information, please contact the event coordinator Ingunn Westerholm on office@oife.org

Save the date – OIFE Topical Meeting “Balancing life with OI” 2023

OIFE Topical Meeting in Stockholm 2023
«Balancing life with OI»

- Topical Meeting & OIFE AGM in Stockholm, Sweden
- Subtopics will include – pain, fatigue, sleep, mobility, family and relationships:
  - June 8 – 11, 2023
  - Collaboration with SFOI
- Venue: Scandic Continental
  - Walking distance from airport express
  - 25 wheelchair accessible rooms
- Target group:
  - People with OI & parents of children with OI
  - Clinicians and researchers
  - Anyone else interested in OI
- Registration will open September 2022

www.oife.org
14TH INTERNATIONAL CONFERENCE
ON OSTEOGENESIS IMPERFECTA

OI2022
30 AUGUST - 2 SEPTEMBER 2022
SHEFFIELD, UK

INCLUDES:
Updates on developments in current scientific research
Medical and surgical treatment options
Related topics such as rehabilitation, quality of life,
pain, mental health and service development

THE MEETING WILL BE OF INTEREST TO:
Metabolic bone specialists
Endocrinologists
Adult physicians
Paediatricians
Dentists
Geneticists
Genetic counsellors
Nephrologists
Neurosurgeons
Orthopaedic surgeons
Nurses
Occupational therapists
Physiotherapists
Psychologists
Radiologists
Molecular biologists
Cell and stem biologists

Please visit the conference website to view the
full programme and for more information

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

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