Unbreakable spirits...or?

By Ingunn Westerheim, OIFE president

The last three months has been a special period of time for OIFE and myself. I will never forget the moment when Eero told me that our vice president Ida Männistö (30) was no longer with us, but had passed away suddenly in her home. To me, Ida was both a colleague in the OIFE Executive Committee, but she was also a friend, a kindred spirit and a young person with OI. Ida was curious, smart, fearless, engaged and never afraid of new challenges. She lived life to the fullest and we all feel very sad that she is not around us anymore.

In dark moments I wonder if people with OI sometimes live too much. As many other people with OI, Ida gave one hundred per cent whether it was work, education, dating, travelling, partying or other life projects. Are we sometimes trying too hard to be "normal" and live our lives to the fullest while our bodies are telling us take some time to relax?
Usually I am able to create some kind of distance between myself and sad things that happen in the OI-community. With several hundred OI-friends on Facebook, I get first hand information anytime someone has a bad fracture, an accident or when somebody dies. This can of course cause bad feelings and worry. But usually I’m able to cope.

But this time it really got to me. I felt physically ill for at least a week after Ida died. I had all kinds of weird physical symptoms and my thoughts were stuck on repeat. What happened? Was it because of OI? Should I also be worried? Are we working too much? Should I take more care of my health? All of these questions were haunting me and I completely lost my ability to concentrate...

And Ida is unfortunately not the only young person with OI who has left us too early lately. One and a half month later we heard about Michael from Denmark. And I know there are others. We definitely need more research and knowledge to understand the causes of these deaths. And we must be able to talk about it, even if it's causing anxiety in the community. Just like we have to be able to address other taboo topics like overprotection, sexuality, obesity, depression, anxiety and so called inspiration porn. Are we really unbreakable spirits or are we just as fragile as anyone else?

I'm not entirely sure how to address all these complex issues. But I do know that we have to address them in an inclusive way without prejudice. Because all people are different! So are people with OI and their friends and families...

Statistically people with OI are good at coping with our rather complex disease. We score the same or even better than the average population in research studies about quality of life - asking about depression, anxiety and coping skills. Why? Are the questions just wrong? Are we hiding something? Exaggerating? Perhaps we just have psychological coping skills we have learned from an early age.

But one thing is certain: We are not invincible and we are certainly not unbreakable - even if we sometimes like to project an image of it! A few weeks ago daredevil Penny Clapcott from the UK tweeted: "I wish the OI community was able to talk more freely about this, but with the slogan "unbreakable spirit" its hard to show that at times we're definitely just as fragile. There is still a lot of work to do around PTSD of breaking the stigma."

Because despite being strong, fierce and having great coping skills - people with OI still face psychological problems like depression, anxiety and even Post Traumatic Stress Disorders (PTSD). But the "system" struggles to care for it, because it has more focus on our physical challenges. In a multidisciplinary setting of OI-management, many will agree that the PT, OT and a medical doctor have a natural place. But what about the psychologists?

Some (but not all) people with OI have psychological issues that are not addressed, which they need professional support to handle. The Quality of Life 4 OI-conference was a first important step in addressing this need, with their deep dive session about this topic. But we need to continue the process started in Amsterdam. This includes information about psychological support in our fact sheets, websites, multidisciplinary clinics and guidelines being developed.

And perhaps the first step is admitting to the fact that we are not unbreakable spirits. People with OI can be strong, persistent, knowledgeable and much more. But we can also break both or bodies and our mind. Or fall into a deep dark hole. And it should not be based on luck if you have someone there to catch you when you fall...
What is the OIFE doing?

By Ingunn Westerheim - OIFE president

It has been a challenging and emotional time after the sudden passing of OIFE’s vice president Ida Mannistö from Finland in October. Ida had not been part of the Executive Committee (EC) of OIFE for more than a year, but she was a natural member of the team and she will be deeply missed.

On the more positive side - 2019 was also a year with many activities for OIFE and we were represented on many arenas, where these were the ones creating most work:

- See, Hear, Smile! OIFE Topical Meeting in Latvia
- Being part of the establishment & ad hoc management committee of the European Rare Bone Forum incl. two network meetings in Salzburg and Amsterdam
- Preparations connected to Quality of life 4 OI-conference in Amsterdam
- Advice from patient perspective in the Asteroid study (Mereo Biopharma)
- C4C - MOI study - advice and recruitment of patient representatives
- Assisting ERN BOND with their registry survey

It’s obvious that our networking and communication work has had an effect, because during 2019 we got an increasing amount of requests from all kinds of stakeholders who want our advice or who want to work with us some way or another. Our challenge is reporting back to the community about all the arenas where we are involved. But we try to do so through our website, social media and the magazine you are currently reading. Follow our blog on our www.oife.org for more recent news and happenings.

See, Hear, Smile!

OIFE’s topical meeting See, Hear, Smile! took place in Riga, Latvia from June 14-15th 2019. It was four years since our last topical meeting in Oslo, and every time we tend to forget how much work it involves gathering more than 80 participants from 22 countries. See, Hear, Smile! brought excellent talks about eyes, ears, teeth and craniofacial issues, great discussions and good opportunities for networking in a relaxed atmosphere. A big thank you to the lecturers who came to Riga and contributed with their knowledge, with little or no compensation!
OIFE Annual General Meeting (AGM)
At the AGM in Riga the Executive Committee of OIFE accounted for OIFE’s activities in the past year. Future plans and policies were discussed and members exchanged news and ideas. Dr. Eva Åström from Sweden gave an update on ERN BOND and the company Mereo Biopharma gave us a status on the Asteroid study and presented opportunities and challenges regarding real world evidence and the need for registries.

ICCBH & ERBF
Shortly after Riga OIFE was represented at the International Conference for Children’s Bone Health from June 22nd - 25th, this year mostly to network with central professionals and people from the rare bone community. OIFE was also one of the central parties behind the establishment of the European Rare Bone Forum (ERBF), which has taken a lot of time and energy in 2019.

Course on registries in Rome
From September 23rd - 27th our honorary member Taco van Welzenis represented OIFE at the International Summer School on Rare Disease Registries and FAIRification of Data in Rome, Italy. It was a part of a series of training activities proposed by the European Joint Programme on Rare Diseases (EJP RD) and was made up of 5 intense days of residential training organized by Istituto Superiore di Sanità (ISS) in close collaboration with other stakeholders.
EURORDIS-meetings
On November 8th Céu Barreiros (treasurer) represented OIFE at the Council of Federations-meeting in Brussels hosted by EURORDIS. Among the topics of the meeting of the 2019-meeting was status on ERNs, funding for research through Horizon Europe, news about the European Union and access to new therapies for people with rare diseases in Europe.

On October 15th OIFE was represented by vice-president Dace Liepina at the 28th EURORDIS Round Table of Companies Workshop in Barcelona – “A New PARADIGM of Meaningful Patient Engagement in the Life Cycle of Medicines”.

Quality of Life 4 OI in Amsterdam
Many people from the OIFE and OI-community in general attended the Quality of Life 4 OI-conference from November 22nd – 25th in Amsterdam. OIFE hosted the deep dive session 5 on patient empowerment together with OIF, BBS and the other organizations. Many people from OIFE contributed in various ways even if the biggest workload was definitely on the foundation (and OIFE member) Care4BrittleBones.

Meetings and events
Looking back it’s fair to say that 2019 was the year of the many meetings. As you can see from the list below, the amount of arenas where someone from OIFE was represented is by far the most expansive during a 6 months period of time. In addition to the meetings below, we have provided input to Mereo Biopharma (Asteroid study) and we’ve been asked to find patient representatives to other research studies. We have also attended many meetings connected to ERN BOND, to Quality of Life 4 OI & Key4OI and to C4C (MOI-study about Losartan). OIFE president also had numerous videocalls (VCs) with OIFE members, volunteers and members of OIFE MAB.
Other meetings

In September OIFE was represented by Ingunn at the annual family conference of the Brittle Bone Society from Sep 13th - 15th and Anna also attended the AGM of ASITOI (Italy) from June 28th - 30th.

These were the most important ones the last 6 months that hasn’t already been mentioned:

• EUORDIS Summer School, Barcelona, Spain, June 10-14th (Claudia Finis)
• MOCA - Mechanism of Coordinated Access to Orphan Medicinal Products, Diemen, The Netherlands June 17th (Margriet Crezee)
• Meetings European Rare Bone Forum (ERBF), Salzburg, Austria, June 23 and Amsterdam, The Netherlands, Nov 22 (IW) + several VCs in ad hoc management group (IW)
• EUORDIS Webinar on registries June 25th (IW and TvW)
• European Medicines Agency, Amsterdam, The Netherlands, July 9th (TvW)
• TC with company IQVIA (on behalf of client) on unmet needs in OI-research, July 22 (IW)
• China Dolls 10th anniversary, Yantai, China, August 3rd - 6th (Oliver Semler - OIFE MAB)
• VC Kara Ayers OIF about PCORI & patient engagement, September 4th (IW)
• VC OIFE, Care4BB & ERN BOND about Qol4OI, Sep 5th (IW, IM & RTS)
• VC Cecilia Götterström BOOSTB4, Sep 6th (IW)
• Videocall Francois Houveyz EUORDIS about pamidronate in Romania, Sep 9th (IW)
• The 14th International Skeletal Dysplasia Society (ISDS) meeting, Oslo, Norway Sep 11th - 14th (Rebecca Tvedt Skarberg in addition to Antonella Forlino who represented OIFE MAB)
• Preconference seminar ISDS for patient organizations, Sep 11th (IW & RTS)
• TC Kyowa Kirin International, Sep 24th and Oct 18th (IW)
• Conference about Health Technology Assessment in Norway, Oslo, Norway Sep 26th (IW)
• Webinar "Successful industry strategies for implementing Federated European Registries" Oct 8th (IW and TvW)
• VC about repurposing of drugs in OI Anthony Hall, Oct 23 (IW)
• Lecture at seminar on quality registries, Oslo, Norway, Nov 11th (IW)
• Working dinner L. Sangiorgi & R. Tvedt Skarberg ERN BOND & OIFE, Dec 17th (IW & TvW)
• Mereo Biopharma meeting about real world evidence, Oslo, Norway, Dec 18th (IW and TvW)
• EUORDIS Leadership School - during the whole 2019 (RTS)

A big thank you to all the volunteers who represented us in the countless meetings in 2019!

**OI is more than Fractures, Surgeries and Pain – „QUALITY OF LIFE 4 OI“ conference**

_By Meriem Ben Jelloun, Germany_

The conference ‘QUALITY OF LIFE 4 OI’ took place in Amsterdam from the 22nd-25th November 2019. It was expected to gather the scientific, research, medical and many proactive individuals with OI themselves or others representing ‘the patient’ side, which in my view it certainly was successful at. The topics covered at the conference were diverse and encompassing the latest research from clinical trials involving young OIers, such as stem cell injections as a treatment option, to the most efficient medical care structures for patients with OI at a global stage coming from Canada and Hong Kong.
I would like to narrow my reporting to the themes of the conference that stood out to me personally. I would not be mistaken to say that it was probably the first time ever that a conference of this scale managed to shed light on one of the most untapped subjects of all, the psychosocial aspects of OI. As we all know, OI comes in all shapes and sizes, its diversity has posed tremendous challenges to medical teams all over the world for years. Increasingly though, as doctors and surgeons thought they got a grip on how to diagnose, how to manage fractures, how to straighten bones and maybe even how to manage pain. Once again, they found themselves challenged and faced with issues that could interfere with the results, they would expect from the medical and therapeutic treatments provided to their patients.

The psychosocial consequences of OI are indeed diverse and very common. These psychological barriers are also very independent of the type of OI and the age of the person with OI. They range from the sense of isolation as fractures and hospital stays reoccur more often than wanted to depression, self-esteem and confidence issues and not to forget the extreme lingering fatigue that interferes with the daily activities and performance of children and adults alike.

There was one workshop titled ‘Psychosocial Aspects of OI’ led by wonderful speakers: Kara Ayers (USA), Claire Hill (UK) and Ute Wallentin (Germany). The workshop allowed for discussions to take place around the topics of fatigue, mental health and pain management. Different coping tools and mechanisms were also suggested. In addition, there was a wonderful exchange of knowledge and ideas between the speakers and the audience which allowed for the OIers to voice what mattered to them and had a say as to what was important and what was not. Though it seemed that we are still right at the beginning, we definitely should not let that discourage the efforts and the meaningful work that has been done so far to tackle these issues individually. All with this in mind, it becomes evident that it is an urgent matter to include a well-structured psychological support as a vital and essential part of any OI treatment starting, preferably, in early childhood. Subsequently, there is an urgent need to improve early screening methods and treatment options to overcome the psychosocial problems for sure.

To sum it all up, I was extremely impressed and glad to see the active involvement of empowered young and adult OIers who are taking the reins of their care and lives to personally direct OI-specific research with the aim to improve the quality of their community in the context of the individuals’ varied perceptions. With that I would call on the conference organizers for a follow-up in few years to see what has become of all the studies presented.
The title and graphic identity of the Latvia meeting Jean Moitry and I attended in Riga is a reference to the Chinese tale of the three monkeys. One of them doesn’t see, the second doesn’t hear and the third hides his mouth. This is an optimistic message because the meaning of the parable is that if you choose not to listen to evil, not to look at it, nor to repeat negative ideas to others, you are a wise-man/wise-woman.

More literally, when one is a person with OI, one may have in addition to orthopedic problems, issues with one’s sight, or one’s hearing, or with jaw deformities or one’s dental system that may cause pain, teeth fragility all the way to affecting one’s self-esteem especially during teenage years (smile). Behind this humorous and welcoming entry, the rich program Ingunn and OIFE had devised contained arduous scientific material not always easy to apprehend for a neophyte like me, when it comes to all the background knowledge about our pathology. Let’s try to summarize and indicate a few tracks.

Introductory words the organizers were followed by a general presentation of OI by Dr. Antonella Forlino, explaining different ways to classify OI, the Sillence classification and the classification with the 19 different genetic types. Research pertaining to the mutation classification could lead to more specific treatments and follow up in the future.

Day one was first devoted to eye pathologies which can occur due to thinner sclera and cornea and then to dental treatments and jaw function problems pertaining to OI patients suffering in these areas. We learnt that serious vision problems such as glaucoma or cataract seem to affect less than 5% of OI people of any type, and are therefore reported as less prevalent in OI patients than in the average population. Dr Janna Waltimo Siren and Dr. Suken Shah then approached skull base deformities and basilar-invagination which may appear in as much as 25% of the OI population (30% of types III and IV). Concerning dentinogenesis imperfecta (=DI), according
to an Eastman Institute study, among 50 patients of all types of pathologies suffering from DI 28% had this condition due to OI. DI mostly affects OI types III and IV, much less type I. In Northern Europe, several studies have been devoted to its various forms notably within the TAKO and the Eastman institutes networks. Dr. Agnes Bloch-Zupan, a pediatric dentist and biologist at the IGBMC Centre of reference on mouth and dental disorders in Strasbourg (France) then spoke of her research within the European program Interreg V RARENET on rare dental diseases, among which OI had been included. That same day we learnt from Dr. Manuel Joaquin de Nova Garcia that the effects of anti-resorptive treatments such as bisphosphonates on the evolution of dentinogenesis have been studied: they seem to be causing dental malocclusions in some patients. Studies about the consequences of such treatments in relation to hearing loss apparently have not yet been made.

On the second day, we were given an overview of the various forms of hearing deficiencies in OI. It is commonly said that about 50% of us will suffer from hearing loss. Within the framework of a Danish study, 60% of examined ears in O.I. patients were affected. A lot was learned about the prevalence of deafness due to transmission failure (middle ear) in comparison with hearing impairment caused by perception defects (i.e. inner ear, cochlea affections). Optimal ways of detecting, treating and following up the various types of deafness in OI, were detailed by audiologist Freya Swinnen.

I noted that there appear to be less studies produced nowadays about “deafness and OI” than in the 1970 to 2000 period. A few existing recent studies however reveal that the most severe hearing losses associated with OI originate not specifically in middle ear — of which the small bones deformations and possible surgical treatments have been explored extensively, as Dr. Pedersen reminded us —, but rather in the inner-ear or in a combination of both conduction and perception problems (i.e. middle and inner-ear deterioration) adding
up with age. As far as we know, “fortunately”, very severe malfunctions and this combination factor occur in less than 5% of OI patients.

The causes of mutations and conditions inducing malfunction in the cochlea in some OI patients whilst not in others have not yet been identified. What we know is that, like the middle-ear bones, a fragile cochlea is likely to suffer from micro-fractures and chaotic bone remodeling. As the temporal bone is also prone to demineralization with age, perceptive cells finding themselves in a hostile environment, begin to atrophy. In the case of severe hearing loss — affecting patient’s capacity to understand spoken conversations even while wearing external hearing aids —, one may have recourse to cochlear implant (=CI) provided one’s cochlea hasn’t been obstructed. Implantation consists in the surgical insertion of a silicone electrode carrier in the inner-ear, coupled with a microprocessor implanted under the skin of the head (see illustration on the right). As surgeon Gunta Sumeraga, Freya and myself emphasized, it is absolutely indispensable to be examined by a surgeon who already knows OI or similar bone pathologies¹ before making the decision to get an implant. With a cochlea wall as thin as ours, risks of cochlear perforation and meningitis are greater, as well as risks that the facial nerve gets stimulated (in addition to the auditory nerve aimed at) when the electrical processor of the implant is turned on, causing facial paralysis or jerks.

We still have some way to go before a solid body of work exists to improve the quality of life of OI people affected by deafness. Although this may be considered a secondary disability because it appears, in its stronger expression, after the age of 40, deafness should be treated as early as possible, when it is still mild. Regular (possibly annual) audiograms should be performed starting in early childhood and throughout adulthood. It is important also to be prescribed and to wear hearing aids as soon as medium range deafness is diagnosed, as well as to get a professional psychological follow up: these are key factors for avoiding the serious psycho-social damage untreated deafness may cause.

¹OIFE can provide a list of such specialist compiled with the help of patients and doctors, and recently enriched by Diane Maroger
Each section of the Riga program began with patient testimonies that had been carefully and sensitively selected by members of the OIFE board, who read them aloud to us. The conference’s audience of about 80 people consisted primarily of medical professionals and secondly of patients and OI family members of all ages, converging from all over Europe and Russia. Such diversity among the attendees surprised Canadian Pr. Jean-Marc Retrouvey who made a rather arduous presentation on 3D modelizations of cranio-facial characteristics in OI. He expected to have a mostly professional audience! However this is very much the specificity of OIFE: mixing audiences to allow active patients to document themselves whilst providing an opportunity for professionals to meet the people who make their practice meaningful, and not just remain among themselves.

The Riga meetings were successful at that, taking place in a beautiful setting and in a very friendly atmosphere. Thumbs up and a BIG thank you to Ingunn, Taco, Ute, Anna, Dace and her husband for their warm welcome. Many thanks also to Zita for her skillful simultaneous transcription that allowed deaf people such as me to follow the conferences.

⇒ A selection of power-point presentations of the individual talks: [https://oife.org/what-we-do/events/topicalmeetings/ - seehearsmile](https://oife.org/what-we-do/events/topicalmeetings/ - seehearsmile)

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**More experiences from Riga**

*By Julien Delaye, OIFE volunteer*

"From Friday 14th of June to Saturday 15th, Riga, Latvia was the place to be for the OI community. On Friday, the sun was shining, temperatures were high, people in the streets wore summer clothes and highly-needed sunglasses, stopping every now and then to appreciate the beauty of the city or some refreshments at a café’s terrasse." Read more about Julien's experiences from See, Hear, Smile! on our webpage: [http://bit.ly/2RY3iVs](http://bit.ly/2RY3iVs)
Impressions from See, Hear, Smile!

**Armaana Ahmad**  
Paediatric Dentistry, Eastman Dental Hospital London

I am a Paediatric dentistry and junior researcher interested in dental anomalies in children with OI. I don’t have a formal relationship with OIFE but would like to!

I think the Topical Meeting was excellent. Well thought out content allowed networking and sharing of current research with experts across the globe. Focussed enough to make every talk highly relevant to people working in similar themes. Personal highlight: Patient led conference, hearing the experiences of patients with OI, seeing the impact of our research and meeting and interacting with people from all backgrounds.

My wish for future meetings: A joint conference looking at teeth in bone conditions - links with Hypophosphatasia and rickets.

**Mengquan Li**  
Dentist from Hongkong University Shenzhen Hospital

I'm a dentist from Hongkong University Shenzhen hospital, Our hospital is the treatment center of south China for OI patients. In our dental department, we received more than 100 OI patients, but only a few received treatment. One of the reason is we don’t have many experience in OI patients. So I came to attend this meeting.

I think this meeting expanded my horizon, and let me know the principle of treatment. I’m interested in Maung Maung Myint and Ann Lindunger's study. We are also puzzled about the long term result of implants. In future, I hope I can hear about more studies about the implants and the BRONJ (Bisphosphonate-Related Osteonecrosis of the Jaw)

**Bruno van Dijk**  
Belgian OIFE-delegate

I’m the chairman of the Belgian (Flemish speaking part) OI organisation. I do have OI type 1 myself. The meeting was very useful and there were many intersections. But the most important for me was meeting other people from all over Europe and see some friends again.

My personal highlight was the part of “hearing”. What I like to be addressed in future meetings? This is a difficult one. Maybe about insurance or legal issues for people with disabilities, what they can claim etc.
I’m Claudia Finis, a psychologist from Germany. I attended EURORDIS summer school on behalf of OIFE in 2019. And indeed it was a school. We had to do a lot of work. Prior to the actual week of face-to-face training we had to read and work on real and invented articles as well as to do online training. The latter consists of several online training modules and is available online for free for anyone to use. So feel free to try it.

We learned a lot about pharmaceutical trials, medical research and how to assess it. Furthermore, we learned how the responsible people come to their decision whether they authorize a drug or not. One way for patient engagement is to work together with the EMA.

**But what is the EMA?**
The European Medicines Agency (EMA) is an agency of the European Union (EU) responsible for the scientific evaluation, supervision and safety monitoring of medicines in the EU. The mission of the EMA is to foster scientific excellence in the evaluation and supervision of medicines, for the benefit of public and animal health in the EU: [http://bit.ly/2RwJSrU](http://bit.ly/2RwJSrU)

**And why is that important for us?**
Because the EMA is responsible for the orphan designation and marketing authorisation. It is responsible for the scientific evaluation of marketing authorisation applications for all orphan medicines in the European Economic Area, as they fall under the mandatory scope of the centralised procedure.

**What is orphan drugs?**
Orphan drugs are medicinal products intended for diagnosis, prevention or treatment of life-threatening or very serious diseases or disorders that are rare. A disease or disorder is defined as rare in Europe when it affects less than 1 in 2,000 citizens. These drugs are called “orphan” because under normal market conditions the pharmaceutical industry has little interest in developing and marketing products intended for only a small number of patients. Pharmaceutical companies can apply for an orphan drug authorization and when granted they will get several benefits. But it’s a long way from the idea to the available medicine. And the work of the EMA goes even further. After authorization the drug is still monitored for several reasons e.g. side effects or long term reactions.
Reporting adverse effects is important
By the way, every patient is allowed or even encouraged to report about adverse effects to the EMA. This means drugs in off-label use, too. This is important and might lead to consequences such as the withdrawal of a drug if needed.

Where and when should patients contribute?
In order to prove that a new drug is safe and efficacious a lot of research has to be done, valid and meaningful data has to be collected. We as patients can contribute along the way. In fact, researchers can be surprisingly ignorant of what really matters to us. For example, a drug that causes measurable changes anywhere in the body but does not improve the issues that are important to us can be completely uninteresting to us and nobody would take it. That would be a waste of time and money for the pharmaceutical company while disappointing for us. So everyone will benefit from good collaboration. There are many opportunities to participate along the way. This graphic gives a brief impression.

Overview of patient involvement along the medicines lifecycle at EMA

Every orange speech bubble means a chance to get involved. In addition patients can give valuable input at a community advisory board (CAB), which is a group of patients who offer their expertise to several public or private sponsors of clinical research.

Research on children is getting more and more important.
In the past, research was mostly done on adult men. That is why there are far less drugs for children. Many drugs are nevertheless used for children, some as off-label use. With varying
degrees of success. Nowadays the EMA tries to test new medicines as far as possible and necessary also for children. This is important because children often react differently. They have a different metabolism, they are still growing and they have different needs. But of course they are not able to participate in a committee. So the Paediatric Committee is probably the only one where not the patients themselves participate but their parents.

Which questions arise when one participates in a committee?
There may be ethical questions and decisions to be made and sometimes the trial has to be modified in order to get valid data. In other cases the research conditions may be so stressful for the participants that a high cancellation rate is to be expected. It looks like sometimes researchers think that it is no problem to attend the trial. But for the family this may mean to drive four times a week many kilometers after school (and work), wait at the hospital for one hour, get treated or examined for ten minutes, drive back all the way but they have to take care to arrive at the hospital before 3 p.m. with a child with special needs. The researchers often don’t take into account that people with a rare disease live far away from the performing university/ hospital and due to their disability they are not able to bare this stress. In addition, the parents may get problems with their employers. No wonder that such trials have a high dropout rate. Here again we can prevent a waste of time and money with our input. What are we willing to do and endure? What is possible, what is not? These researchers have sometimes never seen or talked to a patient with the disease or disability they are researching. They don’t know what it means to live with this disease or disability.

Besides the dry and important work, we also could chat, get to know nice people and do some networking, which of course is also important. And there are these encounters and moments that just delight the heart. I highly recommend the summer school for patient advocates especially for those who are willing to work on a European level.

New OIFE Member: OIF Ghana

By Justina Yiadom-Boakye
President, Osteogenesis Imperfecta Foundation Ghana

Who are you and what is your relationship to OI?
My name is Justina Yiadom-Boakye, a mother to a child living with Osteogenesis Imperfecta. I am also the founder and the current President of OIF-Ghana, who recently became a member of the OIFE.

Can you tell us a few words about Ghana?
Though it might be difficult to give the exact number of people living with the condition here in Ghana, however, using the statistical estimate of one out of every fifteen thousand people ratio, we estimate a total number of about two thousand people living with the condition in Ghana given our total population of about thirty million.
Some of the major challenges facing people living with the condition in Ghana includes the following:

• Bisphosphonate drug Pamidronate is not readily available.
• Mobility aids especially for their small sizes are not readily available and expensive.
• Those we have identified currently have no form of corrective surgeries done for them. As a result of this, most of them have severe limbs and spine deformities.
• Most of the children we have identified have been abandoned by their fathers, living the mothers to care for them on their own.

Can you tell us about your organization?
OIF Ghana was founded on 6th May, 2017. Our main mission is to improve the quality of life of people with OI in Ghana through Education, Awareness Creation, Treatments and Mutual support. We currently have seven OI children we are working with. Most of the people we have identified got to know of us through our limited awareness creation and publicity activities. As a young institution, our biggest challenge has always been funding.

How do you recruit members/promote the organization?
OIF Ghana has lined up a couple of projects it hopes to execute in the nearest future. Some of these projects are listed as follows:

• To organize a large scale publicity campaign in Ghana
• To train representatives’ volunteers across all the 16 regions of Ghana before the end of 2020 to help affected families in the respective regions.
• To pursue for the setup of a medical team that will be in charge of handling OI cases in all the teaching Hospitals in Ghana.
• To have a specialized OI clinic within the Central Regional Teaching Hospital where OI patients will be given proper care and attention every half year.

• We also hope to put up a fund which will be used to assist OI patients financially with surgical operations. This is particularly dear to us because even though we have an orthopedic surgeon here in Ghana that can perform such surgeries, there are no funds to buy rods needed for the operation.

Amongst our great successes, we are the first and only institution in Ghana that has brought OI to the limelight. Though the awareness on OI is very inadequate, we have been able to clear some doubts in the minds of most Ghanaians who supposed OI to be a ‘strange’ sickness. Considering the limited resources available to us, we can confidently refer to this as great success. Also, another success we can boast of is that, all the children we have identified are put on Pamidronate infusion for free with all medical bills born by the organization. We have also provided mobility aids for such persons with the help of our cherished donors.

Plans for the future?
With all that have been said, we look forward to working closely with OIFE to help build and grow our capacities as an infant organization to help give OI the attention it needs most especially in developing countries like Ghana.
Since June 2019, the Netherlands has, for the first time in its history, an unofficial Minister of Disability Issues. Rick Brink who has OI and is using an electric wheelchair, won this appointment during the broadcast of the television programme ‘De minister van Gehandicaptenzaken’ (The Minister of Disability Issues).

Can you tell us a little about yourself?
My name is Rick Brink, I am 34 years old and live in Hardenberg, The Netherlands. When I am not busy working, I enjoy spending time with my family and friends.

How did you get your job as Minister of Disability Affairs?
In June of this year I was elected as the very first Minister of Disability Affairs by the Dutch viewers during a live television show aired by the Dutch broadcasting network KRO NCRV. I applied for the job in March, had to go through three selection rounds and made it through to the live show in June with 5 other candidates. Out of the one-hundred-and-thirty people who applied I became the first Minister of Disability Affairs.

How does a ‘normal’ day look like as Minister?
From the moment I was elected there has not been a normal day. I now represent the needs of the 2 million people with disabilities that live in the Netherlands. So you can imagine we, my team and I, are very busy trying to truly make a difference for all the people we represent. Every Monday we discuss the upcoming week, so I am well prepared. But still every day is different and often unexpected things happen. For instance, I can get a call if I can be a guest on a talk show the next day or do an interview, that wasn’t planned, that makes the evening news. But the majority of my days are spent talking to influential politicians, people with disabilities and organizations and interest groups.

What are your main goals as Minister of Disability Affairs?
My absolute main goal is to create a truly inclusive society and I have three main goals I hope will contribute to creating an inclusive society:

First of all, an inclusive start for children with disabilities, because I believe we can do better. It is my goal to create a society that is accessible to every child. This means a special focus on inclusive education and on inclusive playgrounds where children with and without a disability can play together. I believe when children with and without disabilities interact with each other from early on, this will reduce stigma and negative thoughts and support inclusion.
Secondly, in the Netherlands a lot of students with special needs have trouble finding a suitable internship. This sometimes leads to them not being able to complete their degree. That is why I have set myself the goal of creating thousand internships for students with special needs.

My third and final goal is to promote the visibility of people with special needs in the media.

People with disabilities are rarely visible on Dutch television and when they are, they are usually there to talk about their disability, not to talk about their talents or expertise. I want to change this. I want to focus on their abilities instead of their disabilities. People with an impairment are more than their disability, this is a part of them but not all there is to them. They have talents and it is high time these talents are recognized and seen.

That is why we created De Koffer van Rick. In De Koffer van Rick we collect the CVs of talented people who also happen to have a disability. We will present these CVs to producers to help and encourage them to create more inclusive programs.

**What are the biggest opportunities and challenges of having this position?**

My motto is: We are not our disability, we are our talent! I hope that I, through this position and the fact that I have a disability myself, I can contribute to changing the way people with disabilities are viewed in our society.

**Is there anything you like to tell the readers of OIFE Magazine?**

It is important that people with a disability are open about their situation. Some people recognize the condition I have and know what it entails, but there are many who don’t. In my experience, when people don’t know anything about the condition I have, people can get uncomfortable. They are unsure whether they can ask me questions, don’t know if and when to help me. So I explain the condition to them and how it affects me. By starting the conversation about my situation and how it affects me, people understand it better and are able to offer me the support I need. If you are honest and open about your situation, what you can and cannot do and what you would like others to do for you, you make life easier for yourself as well as for the people around you.
Can you introduce yourself?
My name is Veronica Tulli, stage name LuluRimmel, I live in Rome and I am a singer, songwriter and performer. I sing and perform in several shows of different nature, sometimes also dark and grotesque, this as LuluRimmel, my character: a doll that plays ukulele. I am also part in an acoustic duo with my partner Guido Maurizio Doria, called “Pancake Drawer”. In this project I have the chance to express myself more as a songwriter. We write together the lyrics and the music and this is really the thing I love the most. I sing and play ukulele, and he plays the guitar.

How has OI influenced your arts and you being an artist?
I am pretty sure I would have been an artist even without OI, because I think that arts and music are a need that each one can feel in a different way. From a social point of view, I am sure that my projects have somehow an impact in the world of disabilities, it is like people can read a social and cultural statement in my art. I believe that music is simply just music, and that it has itself a so powerful and strong message to overcome any obstacle and barrier. I sing and play because I love doing it, I do not care so much of being an example, I believe that telling about myself and express myself through music have a meaning for the people that are listening to me or watching me.

Tell us more about “Heart of a Doll”, how did the project and the concept of it begun?
The project was ignited during an informal meeting with some good coffee with Antonio Di Domenico, the director of the documentary. At the beginning it was supposed to be the first episode of a series: people with different backgrounds and different physical difficulties, telling their arts and stories. “Heart of a Doll” talks about my life, mainly the artistic side of it, and highlights the adventures, the losses, the victories...it is all authentic. It is the story of a girl, like many others, following her dream, to be a full-time artist and make a living with it.

Do you have other upcoming projects?
For the time being Guido and I are focusing on our duo “Pancake Drawer”. We are thinking about recording an album, the first one with our own unreleased tracks.
Why did you start singing and acting?
I would refer to myself more as a performer, to be honest. Even in the documentary there is not that much acting, I have attended some acting classes in the past but never at a professional level. About singing, I have been singing since when I started talking. As a kid I was always singing, it made me happy.

Which topics are closer to you?
In my songs I often talk about my dreams and my hidden places. I mostly talk about love. Actually, I believe that every song ever written is somehow a love message. I talk about simple stuff that everyone can relate to and understand. Feelings belong to everyone. I would like that by listening to one of my songs people could say “Oh Yes! It has happened to me as well; I have felt like that!”

What do you prefer between singing and acting?
When I play my character LuluRimmel the acting part is really important, but it wouldn’t mean much without the songs played with ukulele. I have fun playing this role of a doll, a bit silly, but that is never clear if she is really silly or if she fake it, and who laughs with no reason. It allows me to play with the first impression of people, with their prejudices about my disability that is physically quite clear and visible. When I sing and play with my duo, instead, there is no character, I am myself with our songs. Acting is nice and fun, but honestly the hardest and the best thing at the same time is being themselves without masks.

Your most scary or dangerous work experience
There is this place called “Bottega degli Artisti” where I do perform every now and then. Every time I perform there, I end up climbing on some strange and potentially dangerous places. On a piano, on some kind of carts, on a bench, on some boxes, or a library…Due to my short stature it is more spectacular when I am placed somewhere very high. Dressed up as a doll I keep myself still and then I greet by waving my hand to the audience when they come in, as they ask themselves if I am real or fake. Yes, this is or can be somehow dangerous, but I have fun and, for sure I am not afraid nor scared!

What is or should be the role of artists in the society?
An artist has the privilege to leave a mark in the everyday history. The causes he/she supports and the messages that he/she spreads are heard from much more people, especially if he/she is a well-known artist. What is really perfection is when by only expressing himself/herself an artist can bring and spread a message for all, a message of equality among people no matter the diversity.

What is your secret dream?
Play in some New York club with “Pancake Drawer”. I have never been there and I would love to and let people listen to our music and our songs.
OIFE Youth event
By Gareth Cummings, Trustee Brittle Bone Society

The 2019 event was hosted by the Spanish OI Society AHUCE and saw 49 participants between the ages of 18 to 35 from all over Europe (10 different countries in total) come together for a variety of activities and socialising. Gareth Cumming from the UK attended the event and here is what he wrote:

Getting There
This was my first time visiting Bilbao and due to limited flights I had to fly the day before giving me the opportunity to scope out the city before the others arrived.

I was a bit apprehensive at first due to the fact that Bilbao is situated on a very mountainous area meaning that it was very hilly and difficult to get around by manual wheelchair. The weather also didn’t help as Bilbao is located in the very north of Spain and combined with the time of year meant that the first few days brought torrential rain – worse than British weather!

Day One
On the first day of the youth event I arrived at the BBK Bilbao Good Hostel. It was chosen as it is a hostel which has been designed to cater for many disabled guests, and it was the perfect venue to cope with all 25 wheelchairs that attended. Due to everyone arriving at different times from different countries there was only time for socialising and getting to know each other through ice-breaker activities.
Day Two
The rain had eased off a little, so we all made our way to the city centre. To get there we had to get the metro train which was a short walk from the hostel and was fully accessible however it did involve going in several lifts which was only big enough for 2 wheelchairs at a time, so with 25 wheelchairs this did take a while! Once we all arrived in the city centre the rain was too heavy for the free city tour that was organised so this gave us all free time to explore the city’s shops and bars and sample the local Pintxos – the region’s version of Tapas. Very tasty! When we returned to the hostel the group collected for the first organised activity, a very in-depth talk about relationships & sexuality.

Day Three
The sun had started to come out and we went on our next activity, a visit to the world famous Guggenheim Museum. To get there we all boarded a large wheelchair accessible coach – which again took forever to load 25 wheelchairs on and get strapped in. Once at the museum we split up in to 3 groups for a guided tour around the museum and talk about some of the museum’s most famous pieces of art. The works ranged from very traditional paintings from artists such as Van Gogh and Monet to a giant metal spider sculpture and a giant puppy made out of flowers. Once the museum visit was over most of us decided to make our own way back to the hostel as loading the bus took too long, so groups of us made our way to the metro station with several stops at the local bars to get some more Pintxos (Tapas) and Cervezas (beer), the evening was finished off by more “socialising” on the hostels roof balcony.
Day Four
The Spanish sun had finally come out to play so we all made our way to a nearby park where we all took part in the “Gymkana”; we all split up into teams and took part in team activities such as quizzes, riddles and physical challenges, ending with a picnic on the grass where it was warm enough to strip off and top up our tans. In the afternoon the majority of us went back into the city centre where we were able to go on the free city tour that had been rearranged from earlier in the week. We were able to take in the amazing views of the city and hear about its fascinating history. The evening culminated in the main party night where the organisers had reserved a local night club where we partied until the small hours. It may not have been wise to hold the main party on the last night as many people had early flights the next day and we all had to check out of the hotel by 10:30am!

Goodbyes
The last day was just enough time for early morning goodbyes through teary eyes and painful hangovers, which was an encouraging sign that everyone thoroughly enjoyed the week and that it was really well arranged and executed by the organisers and their volunteer helpers. The whole trip was fantastic, even with the dodgy weather, and there was only one OI related injury. Unfortunately, being 35, this was the last year that I was young enough to qualify to attend, which is a shame as 2020 will be somewhere in Poland, therefore I would encourage anyone reading this with OI between the age of 18 – 35 to sign up as soon as it’s advertised.
Marie’s Youth Corner
By Marie Holm Laursen, OIFE youth coordinator

I attended in the QualityofLife4OI conference in Amsterdam in November. I wasn’t sure if it was a good idea to go because I had to move apartment two days after the conference. Luckily I decided to go and it was such an amazing trip and I managed to pack my apartment down afterward so I was ready to move.

It was so good to see all the people I already knew but also to meet a lot of new faces. I got new friends at the conference and that is what I love the most about having a disability that you get friends from all over the world. Hopefully next year we will all meet in Poland where the next OIFE youth event will take place. It is going to be so much fun!

At the conference there were so many interesting workshops and speeches. It was just so cool to see how many people who are passionate about improving Quality of life for people with OI in one way or another. At Saturday there were 5 different workshops and you had to choose one of them and it was so difficult to choose because they all sounded so relevant. I decided to hear more about medical treatments and it was very interesting to hear about. We have already come along way in treatments but just to know that we are constantly getting better and finding new ways to help people with OI is so cool.

This is a picture of Stephanie and me from the last night at the conference where there was a dinner party. There were different performances and a guy named Sparsh was singing. He was so good at singing and it was a perfect ending on a good conference.

Founder of Help for OI (HOI) has passed away
Written by Ute Wallentin

We bring you the sad news that Hinke Panjer founder of our associate member organization (the Dutch charitable foundation Help OI - HOI) passed away on October 24th 2019, at the age of 70 years. Hinke had no easy life, but she developed a great passion to help other people living with OI. HOI’s purpose was to support OI individuals all over the world for their medical treatment, with necessary equipment, surgeries etc. Hinke and her partner Harm started the foundation in 1996 and together with several local friends and supporters in her home town they collected thousands of Euros over the years for medical equipment to Georgia, wheelchairs and small walkers to Sri Lanka, Ecuador, the Philippines, Nepal, Russia and Cuba. HOI as an organization has now been officially closed. You can read a longer obituary on OIFE’s webpage.
Search for professionals & patients organizations who are interested in breathing and sleep management in OI

I am the P.I. of the project “Eat, Breathe, Sleep with OI” financed by Care4BB and As.It.O.I. where we are analyzing the nutritional status, breathing and the quality of sleep in adult OI patients.

Our preliminary results of the ongoing project showed that 13 out of 27 patients need nocturnal mechanical ventilation, 11 of whom did not know it, because of important oxygen desaturation due to obstructive sleep apnoea (OSA). OSA has an important impact on the quality of life since it can be associated with symptoms during the daytime like excessive sleepiness, decreased cognitive functions and fatigue, which may be present for years or even decades without identification.

OSA seems to be a distinctive feature of OI that is easily left undetected since there is no awareness among OIers and clinicians. For this reason, and also because respiratory diseases are the main risk of death for OI (Folkestad et al. Journal of bone and mineral research 2016, McAllion SJ et al. J Clin Pathol. 1996), I am strongly convinced that a consensus statement on breathing and sleep management in OI is now urged and mandatory in order to minimize all the relative problems that strongly impact on the quality of life.

I kindly ask you to help me to put together a group of experts (by offering yourself or indicating the name of colleagues or doctors in your network that can be involved, typically pneumologists and/or sleep experts). Contact: Antonella Lo Mauro antonella.lomauro@polimi.it

Key4OI:
An outcome measurement set

For the last 2 years experts from many different countries have been working together in Project "Key4OI". The aim of this project was to develop a core outcome set for OI, children and adults.

The focus is on:

Outcomes that are relevant for most people with OI, worldwide, throughout a lifetime.
Input was provided through 16 OI focus groups. Their input has been carefully analysed and discussed by the international Key4OI Expert group in bi-weekly meetings and several so called Delphi surveys. And now it is here: The Core Outcome set for multidisciplinary care and research. Check out the animated video summarising Key4OI here: http://bit.ly/2voWys1

In 2020 health care providers in six countries will pilot Key4OI. It will then become available for everyone in 2021. The aspiration of Care4BB is to use Key4OI as a platform for learning about OI together better and faster, eventually supported through big data. Watch that - very exciting - space!

If you are interested to be more closely involved with Key4OI and consider using it in 2021 and beyond, you can become part of the Key4OI - Affiliates group, who is invited for quarterly videoconferences.

To join this group, please email Dagmar Mekking at: dagmar.mekking@care4brittlebones.org

Searching Flying OI Experts for project in Nigeria

The Nigerian OI-organization is very interested in hosting an educational event in 2020 where an experienced OI-surgeon can be invited to come and teach rodding surgery in Nigeria under the Flying OI Experts programme.

Would you consider to volunteer for such a project? Or do you know an experienced OI-surgeon who might be interested in helping? Please contact the OIFE as soon as possible on office@oife.org
ASTEROID STUDY
Mereo Biopharma has announced the results of their Phase 2b study for setrusumab in adults, designed to evaluate increase in bone density measured by High-Resolution peripheral Quantitative Computed Tomography (HRpQCT). More well-recognised techniques were used for the secondary endpoints, which included the traditional and well-established DXA scan, measuring bone density; as well as fractures and quality of life, amongst other aspects of OI. The study enrolled 112 patients in the US and Europe into 3 blinded dose-ranging arms and an open-label arm at the top dose. For information about results you can read press releases from the company here:


C4C - MOI-study is terminated!
OIFE has been working closely with dr. Nick Bishop and the MOI-study team under the Conect4Children-programme, to recruit patient representatives for this project which aim was to start recruiting spring 2020. However late December 2019 the C4C decided to withdraw the funding because there was delivery problems with the drug Losartan, which was going to be tested on children with OI. The MOI-study has therefore been terminated.

NIH-FUNDING FOR OI-RESEARCH
The National Institutes of Health (NIH) in the USA has awarded 31 million USD to the Rare Diseases Clinical Research Network (RDCRN), where the Brittle Bone Disorders Consortium (BBDC) is included. Read more about it here: https://bit.ly/38bDWKb

NEW PHD ON OI
Congratulations to member of OIFE’s Medical Advisory Board - Dr. Lida Zhytnik for finishing her PhD «Inter- and intrafamilial diversity based on genotype and phenotype correlations of Osteogenesis Imperfecta». Her research is based on genetic analysis of large groups of people with OI in Estonia, Ukraine and Vietnam. The PhD is open access and you can read it here: https://bit.ly/2pw5zMX

BONE SCHOOL
Registration is open for the ICCBH Bone School, which is a new 3-day residential international educational course giving the opportunity for participants to learn about and discuss mechanisms, clinical diagnosis and treatment of paediatric bone health and rare bone diseases. The 2020 course takes place at the St Virgil Conference Centre in Salzburg, Austria, on 29 June to 1 July. See https://www.iccbhboneschool.org
WEB BASED RARE BONE CLINICS
The OI Foundation and the Rare Bone Disease Alliance are hosting the Rare Bone Disease TeleECHO Clinic Series first Thursday of every month. Professionals from outside the USA can also join! For more information or to register, visit www.oif.org/ECHO

NEWS FROM SHRINERS
One of the doctoral students at Shriners Hospitals for Children Canada doing research in nursing sciences, Aimée Castro, has just published "Exploring the Views of Osteogenesis Imperfecta Caregivers on Internet-Based Technologies: Qualitative Descriptive Study", in the Journal of Medical Internet Research.

The article intended for parents of children living with OI, can help them understand how technological resources, the Internet, social media, digital platforms, etc., can support and meet the needs of their child. Read the article here: http://dx.doi.org/10.2196/15924

NEWS MEMBERS ERN BOND
The call for healthcare providers to join the 24 existing European Reference Networks closed in December 2019. A total of 841 applications for ERN membership were received. 29 applications were submitted for becoming a member of the ERN for rare bone diseases (ERN BOND). Please check the latest updates for the applicants on the EU Commission website: https://bit.ly/2Ocbe4B

FUNDING FOR EuRR-BONE
We’re happy to announce that ERN BOND has secured EU funding to develop a new registry EuRR-Bone –European Registry for Rare Bone and Mineral Conditions.

Read more here: http://ernbond.eu/2020/01/27/eurr-bone/

PCORI GRANT TO THE OIF
In 2019 the Patient-Centered Outcomes Research Institute (PCORI) awarded the OI Foundation (US) nearly $250,000 to create a multi-pronged engagement strategy to help patients and caregivers become more involved in the research process. To learn more about the project, please visit: https://oif.org/pcor

MEETING WITH FDA
The European Medicines Agency (EMA) is an agency of the European Union in charge of the evaluation and supervision of medicinal products. Their counterpart in the USA is the The Food and Drug Administration (FDA). On September 17th the OI Foundation had a meeting with them to educate them about OI. The OIFE is looking into possibilities of having something similar in Europe as well. Read about the OIF-meeting here: https://oif.org/fdalistening/
EVENT FUNDING AVAILABLE
The European Joint Programme on Rare Diseases has launched a new Networking Support Scheme to provide financial support to applicants for the organisation of workshops or conferences for new or existing/expanding research networks to strengthen collaborations and to enable exchange of knowledge on rare diseases and rare cancers. Patient advocacy organisations, healthcare professionals and researchers are invited to submit a proposal. See https://bit.ly/2Nuy5Ys

PODCAST WITH UTE!
During the QualityofLife4OI-conference the organizers produced a number of podcasts with central people in the OI-community. And who is more central than our very own Ute Wallentin, Honorary Member of OIFE and our Social Network Coordinator? Listen to the podcast here: https://bit.ly/2QWGOow

CHINA DOLLS IN THE US
Exchanging ideas and experiences across borders and cultures is important. December 18th the China-Dolls Center for Rare Disorders visited the OIF the USA to get to know each other better.

OIF 50 IN 2020
Did you know that our friends and partners in the OI Foundation (US) are celebrating their 50th anniversary in July 2020? Read more about the event here: https://oif.org/conference/

GOOD NEWS FROM UKRAINE!
Good news from the Ukraine. The OI-organization Osteogenesis Imperfecta Ukraine (Всеукраїнська асоціація кришталевих людей) was finally successful and the government does now import and finance telescopic rods for children with OI!

OI IN KAZAKHSTAN
Dr. Natalia Belova is a member of OIFE’s medical advisory board (MAB). Recently she was invited by the Ministry of Health in Kazakhstan to teach local doctors about OI and international standards of treatment. Dr. Belova gave lectures, examined patients and took part in discussions about organizational challenges and access to treatment. Children from different cities of Kazakhstan and their parents participated in the educational event.

CELEBRATE
2020 OIF National Conference ★ Omaha, NE
RAISING DION ON NETFLIX
Did you follow the first season of the Netflix series Raising Dion starring Sammi Haney as the main character's best friend Esperanza (who also has OI)? Now Netflix has decided to make a 2nd season. We can't wait to see it! What about you?

ARE YOU BETWEEN 18 AND 30?
Then the European Patients' Forum is looking for you! Applications are now open for their Summer Training Course for young patient advocates. Travel & accommodation costs are covered by EPF. Read more here: https://bit.ly/2QXWoA9

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

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

For more information and a complete list of trial locations visit the website www.BOOSTB4.eu. If you have any additional questions, please contact us via the email address BOOSTB4@clintec.ki.se.
Mereo analysing ASTEROID study, including results from first trial using HRpQCT as a measurement of bone microarchitecture in people with OI

Mereo has completed the first part of its Asteroid trial into setrsumab as a potential therapy in adults with OI and has released the initial results, before continuing to the next phase of the research.

Mereo would like to thank everyone from the OI community who participated in the study, which has yielded some extremely valuable data. The imaging and biomarker data are promising; and we are looking forward to moving forward with our development programme.

Mereo believe that the various different imaging results from the trial will not only support our continued development programme, but also give other researchers useful information that can contribute to future studies into OI and other bone-related conditions.

In particular, we hope that the data will contribute to improving the understanding of OI in terms of bone microarchitecture, particularly around the value of using High-Resolution peripheral Quantitative Computed Tomography (HRp-QCT) imaging technology as a way to measure bone density as an endpoint in OI; and possibly also as a tool in clinical management. We were pleased to have the opportunity to be the first to explore the use of this cutting-edge technology in an OI trial for an investigational medicine, which has never been done before. Mereo remains committed to continuing to research and develop a potential therapy for the treatment of OI and we are looking forward to continuing to work together with researchers, physicians and, most notably, the OIFE and its members.

Disclaimer:
The OIFE is not involved in the design or management of the research studies on page 31 and 32, 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.
### Calendar OI-events 2020

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
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<tr>
<td>March 27th-29th</td>
<td>XXV Congreso nacional sobre osteogénesis imperfecta, Granada, Spain</td>
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<tr>
<td>April 4th</td>
<td>AOI Journées Nationales 2020, Montpellier, France</td>
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<td>April 18th-19th</td>
<td>AGM &amp; Family Meeting DFOI, Copenhagen, Denmark</td>
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<td>April 25th-26th</td>
<td>Annual Meeting SVOI, TBA, Switzerland</td>
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<tr>
<td>May 1st-3rd</td>
<td>NFOI AGM &amp; Family Meeting, Oslo, Norway</td>
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<td>May 8th-10th</td>
<td>OIFE Annual General Meeting, Valencia, Spain</td>
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<td>June 5th-6th</td>
<td>Annual Meeting ASITOI, Rome, Italy</td>
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<td>July 4-11th</td>
<td>Summer camp DFOI, Stevns, Denmark</td>
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<td>July 9-12th</td>
<td>OIF National Conference &amp; 50th anniversary, Omaha, Nebraska, US</td>
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<td>August 29th-Sep 1st</td>
<td>OIFE Youth Event, Krynica Morska (Gdansk), Poland</td>
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<tr>
<td>September 4th – 6th</td>
<td>OI Austria Annual Meeting, Stubenberg, Austria</td>
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### Calendar scientific events & conferences

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<th>Date</th>
<th>Event</th>
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<tr>
<td>February 19th</td>
<td>29th EURORDIS Round Table of Companies Workshop, Brussels, Belgium</td>
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<tr>
<td>April 1st – 3rd</td>
<td>OIF Investigator Meeting, Chicago, US</td>
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<td>April 1st – 4th</td>
<td>39th EPOS Annual Meeting, Porto, Portugal</td>
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<tr>
<td>May 15th – 16th</td>
<td>10th European Conference on Rare Diseases and Orphan Products (ECRD), Stockholm, Sweden</td>
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<td>May 16th – 19th</td>
<td>ECTS Congress 2020, Marseille, France</td>
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<td>June 29th – July 1st</td>
<td>ICCBH Bone School, Salzburg, Austria</td>
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<td>September 5th – 8th</td>
<td>OI2020: 14th International conference on OI, Sheffield, UK</td>
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<td>September 10th – 12th</td>
<td>European Society for Paediatric Endocrinology (ESPE 2019), Liverpool, UK</td>
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<tr>
<td>September 11th – 14th</td>
<td>ASMBR Annual Meeting, Seattle, WA, USA</td>
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### Contact

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