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Editorial

By Ingunn Westerheim, OIFE President

The Voice of People with OI

Welcome to the first special edition of OIFE Magazine! The topic is research and I could have called this editorial "The patients' voice". But to be honest, I'm not too fond of the term patients. Patients belong in hospitals. The OIFE represents people with OI and families, who most of the time live active and meaningful lives outside hospital walls.

There are however many ongoing initiatives that include patients in one way or another: Patient involvement in research, patient centred outcome measures, patient reported data, patient priorities in research and I could go on and on. Many stakeholders want to hear our voice these days. Both because they are interested, but also to secure funding or to get approval from authorities.

Some studies are putting the patients to work, by using data that patients are providing themselves. Both the RUDY study and Rare Commons are mostly based on patient reported data. And, why not? We are the real experts, right? Yes, but not always. If terminology is not understood before the questions are answered online, results can be imprecise and sometimes completely misleading. The Adult History Study from the US showed for instance, that many don't know or understand their own type of OI. And using severity doesn't necessarily help. A person with mild OI who has severe pain, or who has become deaf, might regard his OI as more severe than an otherwise healthy wheelchair user with OI type III.

"By engaging the end users of study results throughout the research process, we are more likely to focus on asking the right questions, study the outcomes that matter most to patients, and produce the useful and relevant results that are more likely to be used in practice."

Patient-Centred Outcome Research Institute (PCORI)

So why is our voice important? The most known impact of OI is fractures. But there can also be wide-ranging systemic, psychological, emotional and social effects of both the diagnosis and its treatment. Objective tests or examinations might be inadequate to understand the impact of OI on a person's health, function or quality of life. This can lead to a mismatch between the person's priorities and perspective and the efforts of clinicians and other stakeholders.

The 'patient voice' can contribute in priority setting as well as influencing health policy in a wider perspective. The patient organisations can encourage clinicians and researchers to do more research in OI. And some foundations provide grants, like Care 4 Brittle Bones and Fundacion Ahuce. As an umbrella, OIFE can contribute with advice, because of our

overview and big network. We can also create arenas for connecting and getting different stakeholders together.

So what does the voice of the patients say? I'm pretty sure it depends on whom you ask. The father of three years old Marko with OI type III from Croatia, will give you a totally different answer than Olga 54 with type V from Estonia. And Peter with type I who has recently become deaf, will have other priorities than Martha, with a femur fracture that won't heal.

How do we make sure that the organisations really represent the voice of people with OI? The simple answer is that we have to work together. The OIFE member organizations need to be informed about the new trends regarding patient involvement. Tools must be developed to gather opinions from the community, like focus groups, queries, panels and webinars. We also need to educate patient experts with OI to meet the constantly increasing need for patient voices in research projects, in European Reference Networks (ERNs) and in consultations with health authorities and policy makers on both national and international level.

"Patient involvement in research needs to be taken to the next level. Rare Disease Day 2018 offers participants the opportunity to be part of a global call on policy makers, researchers, companies and healthcare professionals to increasingly and more effectively involve patients in rare disease research." - EURORDIS

Rare Disease Day 2018 took place one month ago and the topic was research. Some of you were perhaps attending national RDD events. OIFE was represented at the European Parliament in Brussels where BOND-ERN's launched a report on diagnostic challenges in OI. No matter what you did – I hope you had an interesting Rare Disease Day. As you all know - it's only a warm up to Wishbone Day!

Rare greetings from Ingunn
OIFE president

What is the OIFE doing?

We had our annual Executive Committee (EC) meeting in Milan last week-end. We had two almost whole days to discuss topics more in depth, something which is really needed at the moment. I hope you will enjoy this second edition of this extended version of the OIFE Magazine, with the same main topic as Rare Disease Day 2018 - research.

Meetings and events

In addition to many meetings with OIFE member organisations and volunteers, the OIFE has organised or attended meetings with the following stakeholders the last 3 months:

- EC Skype January 16th and Feb 20th
- Week-end EC-meeting Milan, Italy March 9-10
- EURORDIS-webinar about recruiting & managing volunteers Jan 17th (IW)
- Skype with dr. Kassim Javaid about registries & BOND-ERN Jan 25th (IW)
- Meeting with BOND ePAG Rebecca Tvedt Skarberg Feb 6th (IW)
- Skype with OIF & Shriners Montreal about research project Feb 13th (IW)
- ECTS-webinar - about research opportunities in BOND Feb 8th (IW)
- Skype Claire Hill (member of OIFE MAB) Feb 9th (IW)
- Skype Mereo Biopharma, March 6 (IW)
- Establishment of Swedish OI-organization in Stockholm, Sweden, March 17 (IW)
- National conference on medical quality registries, March 20-21 (IW)
- Skype UCB (pharma company), March 23rd (IW)



OIFE AGM 2018 - "Back to the Future"

The BBS turns 50 years in 2018 and we will celebrate OIFE's 25th anniversary & Annual General Meeting (AGM) at the same time. OIFE AGM will start Sunday morning and last until lunch Monday 20th of August. Do you have photos or documents from the OIFE history? Please help us out by sending an e-mail to office@oife.org

Rare Barometer Voices - can OI become the strongest voice?



OIFE is encouraging all individuals with OI and parents of children with OI to register at **Rare Barometer Voices**, because it can give us important data we can use for our policy work. If enough people with OI answer the surveys from Rare Barometer Voices, the OIFE will get access to aggregated data for OI specifically and for patients within ERN-BOND. This will make it easier for us to voice the needs of people with OI in our policy work. If enough people with OI from one country answer - the national organisation can access the data for people with OI in their country. Please encourage your members and patients to register here: <https://www.eurordis.org/voices>

ERN-BOND & OIFE

The OIFE has developed our collaboration with the European Reference Network for Rare Bone Diseases (BOND) further. We've had discussions with working group 5 (databases) about how we can collect information about existing databases/registries and initiatives in the OI-community. We've also been communicating with working group 4 (clinical trials) about how we can gather the patients' priorities in research.

On 28 February 2018, OIFE attended an ERN-BOND event at the EU Parliament in Brussels, Belgium. Read more about this under News from BOND!

The OIFE is worried about the consequences Brexit might have on ERN-BOND, since around 30 % of the healthcare providers in ERN-BOND come from the UK. Several of the working group leads in ERN-BOND are also from the UK. OIFE therefore supports the statement from EURORDIS calling on the European Commission to secure the continuous and sustained involvement of UK healthcare providers as members of European Reference Networks (ERNs) after Brexit. Please share and support it widely in your networks and on social media: <http://bit.ly/2tvJ7p9>

Are you interested in joining the group of patient representatives who receive more extensive information about BOND through Facebook Workspace? Then please contact president@oife.org

OIFE partner in new Shriners research project

The OIFE has entered into a partnership with the OIF and Shriners Hospital in Montreal on the research study: 'A Global Assessment of the Out-of-Pockets Expenses Incurred by Families of Children with Osteogenesis Imperfecta ("Costs in OI")'. OIFE's member organisations worldwide will get the opportunity to be involved. The purpose of the study is to uncover the costs that OI inflicts on individuals and families. Our hope is that more knowledge will help our organisations both on a national and international level to improve our policy work.

OIFE passport now in Hungarian and Latvian

Thanks to volunteers from Hungary and Latvia, we now have our OIFE passport available to download as a PDF from our webpage both in Hungarian and Latvian language. <https://oife.org/en/documents/oife-pass>

Medical Advisory Board of OIFE is now online

Since last magazine, Claire Hill from the UK has joined OIFE's Medical Advisory Board. She will represent the OIFE at the OI Foundation's investigator meeting in Chicago in April, where the main topic is outcome measures in OI. You can now download a list of the members of OIFE's Medical Advisory Board from our webpage: <https://oife.org/en/oife/medical-advisory-board>

Questions? Please e-mail president@oife.org



News from ERN-BOND

*Interview with dr. Luca Sangiorgi (Coordinator ERN-BOND)
& Mathias de la Calle (Project Manager ERN-BOND)*

The poster features logos for the European Reference Network, ERN, FEB 28 2018, the European Parliament, and S&D. The main title is 'EUROPEAN REFERENCE NETWORKS ACCELERATING AND IMPROVING DIAGNOSIS FOR RARE DISEASES PATIENTS'. Below this is the subtitle 'An ERN-BOND White paper on Diagnosis'. The event is hosted by MEP Elena Gentile (S&D, IT) and features a keynote speaker, Vytėnė Andriukaitis, Commissioner for Health and Food Safety. The event took place at the European Parliament, Brussels, Room ASP3G2, on Wednesday, 28 February 2018, from 13:00 to 15:00. A list of participants includes Inês Alves, Meena Balasubramanian, Marcello Bufo, Valérie Cormier-Daire, Josep Figueras, Yann Le Cam, Luca Sangiorgi, Maurizio Scarpa, and Martin Seychell. The event is held under the high patronage of the European Parliament.

On the occasion of Rare Disease Day (February 28th) the European Reference Network for Rare Bone Disorders (ERN-BOND) organized a European Parliamentary Meeting called: **European Reference Networks – Accelerating and Improving Diagnosis for Rare Diseases Patients**. The meeting was kindly hosted by MEP Elena Gentile (S&D, IT) with support from EURORDIS. The OIFE was represented at the event by Belgian OIFE-delegate Filip De Gruytere as well as Dagmar Mekking from supporting member Care 4 Brittle Bones.

The aim of the event was to present the findings of the ERN-BOND White Paper on Diagnosis and discuss solutions on how to best reduce the average time to accurate diagnosis for rare bone diseases. The event also marked the first successful year of the 24 ERNs now operating across Europe.

Dr. Vytėnė Andriukaitis - European Commissioner for Health and Food Safety, was one of the invited speakers. He expressed gratitude to the work that the work ERNs are doing and highlighted three challenges for the future of the ERN:

1. *to ensure the integration of ERNs into national or regional healthcare systems*
2. *to ensure full support to the ERNs*
3. *the selection and integration of new members in the 24 current ERNs*



Which countries are included in ERN-BOND?

Countries currently represented in ERN BOND are Belgium, Czech Republic, Estonia, France, Germany, Italy, Netherlands, Portugal, Sweden and UK. We share a common goal with OIFE that OI-organizations in countries included in BOND should be in touch with the involved healthcare providers in ERN-BOND.

Regarding your question on including new healthcare providers from more countries - this is a question for health authorities in the member states.

UK has many healthcare providers in ERN-BOND Are you worried about Brexit?

Yes, as one of the ERN Coordinators, we've sent a letter to The Directorate-General for Health and Food Safety affirming it'd be a relevant loss if health care providers (HCP) from UK couldn't be part of ERN BOND. A statement from the coordinators of the 24 ERNs can be downloaded here:

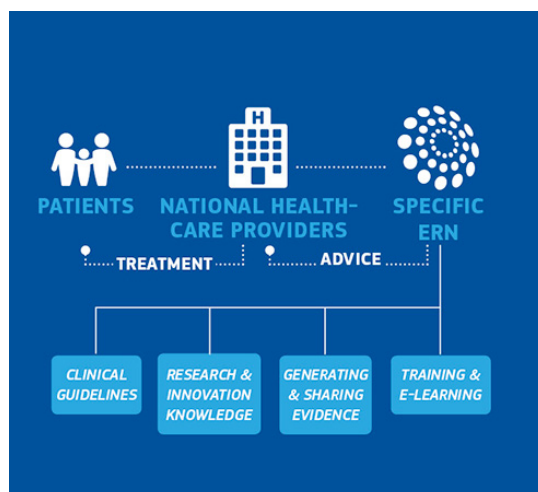
<https://bit.ly/2Gv1iRH>



What are the most important activities in BOND the coming year?

Our most important activities include:

- Increasing the number of patients consulted through the Clinical Patient Management System (CPMS) and to assess strengths and weaknesses of the system;
- To produce draft guidelines for Osteogenesis Imperfecta (as our pilot diagnosis)
- To organize an international consensus meeting introducing the proposed guidelines for OI to start the process of pathway harmonization;
- To map the research activities and expertise within BOND partners through a survey, to facilitate collaborations opportunities for European funding
- To start a discussion with private companies and try to start a clinical trial in 2019, in line with the policies delivered by the Board of ERN Coordinators and European Commission.
- To develop a specification for a rare bone disease registry to improve our understanding of the natural history of rare bone diseases, combining all clinical data into the registry of registries
- To organize a face-to-face meeting with ERN Coordinators to start a platform between different ERNs (inter-ERN), starting with the diagnose of hyperphosphatemia.



EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES

Share. Care. Cure.



European
Reference
Networks



European
Commission | Health

What is the CMPS?

On 20th November 2017, the Clinical Patient Management System (CPMS) was released. A pilot phase lasted until end of February 2018. CMPS is a secure web-based application to support ERNs in the diagnosis and treatment of rare or low prevalence complex diseases or conditions across national borders. The data model running onto the CPMS conforms with European standards, nomenclatures and strategies to share health data syntactically and semantically in the Health sector.



How do we make sure that all relevant stakeholders are involved in ERN-BOND?

The ePAGs (patient advocacy representatives) have a relevant role, since they are involved in all the working groups of BOND. Our network is planning to participate in the following events related to patient involvement - World Health Day, Rare Disease Day, Osteogenesis Imperfecta Awareness Week - May and Dwarfism (Achondroplasia) Awareness Month.

Do you have a message to the readers of OIFE Magazine?

The central role of patients and their families in contributing to BOND activities is not only a statement. We believe that the active participation of OIFE was precious for the first year of BOND and shall increase in the next years.

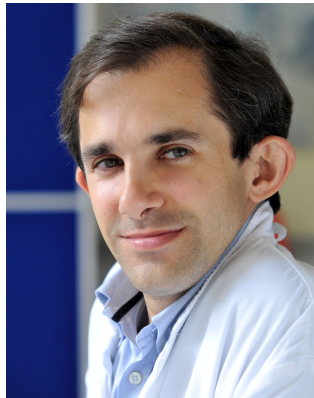
A concrete contribution for the coming year could be the collaboration for the development of studies on quality of life and the definition of outcomes for therapies, with real and clear impact for the patients.



Who is Who? OIFE's Medical Advisory Board

Oliver Semler

My name is Oliver Semler. I am a paediatrician at the university hospital in Cologne, Germany. I am responsible for the centre for rare skeletal diseases in childhood where the OI outpatient department is affiliated. In Germany we are the centre with the most patients and treat more than 200 children with OI. We offer a broad therapeutic concept including medical treatment (bisphosphonates), surgical treatment for fractures and deformities with telescopic rods and we are connected to a rehabilitation centre.



My professional involvement in OI started 15 years ago when I moved to Cologne to work in this outpatient department. Because I am affected by OI myself, I am involved in the national patient organisation since many years and have a lifelong experience in OI. My job is to counsel families regarding therapeutic strategies and to provide medical treatment if needed and to take care of the families while they stay in our hospital or in our rehabilitation centre. Besides treating the individual patient I am involved in research to improve treatment of OI people in the future.

Regarding the most important goal/task of the OIFE - I think we have to bring into focus the improvement of quality of life for OI patients which is not necessarily directly related to laboratory and bone mineral density measurements. Also I think the acceptance of disabled people in the society will be a major political issue in just a few years. Especially keeping new prenatal diagnostic possibilities in mind, which will allow a diagnosis very early during pregnancy.

This topic can only be dealt with by strong organisations and I wish that OIFE could develop some hints for the national organisations so we can combine our efforts.

Antonella Forlino

I am Antonella Forlino. I obtained my degree in Biology in 1991, my PhD in Biochemistry in 1994 and my Specialization in Genetics 1997 at the University of Pavia in Italy. I am currently an Associate Professor of Biochemistry at the Department of Molecular Medicine, Biochemistry Unit of the University of Pavia.



I started to be involved in basic research on OI during my Master Degree Thesis project in 1990 and since then I kept being involved on studies related to OI. I have a long-standing and very stimulating relationship with the Italian OI-organisation (ASITOI) since I was a master degree student and I am now member of the ASITOI Scientific Committee.

My research activity is almost exclusively focused on the molecular, biochemical, and functional study of Osteogenesis Imperfecta (OI) in its dominant and recessive forms with a great interest in translational studies. In December 2010 I started a new project focused on the use of zebrafish as model to better clarify OI pathophysiology and develop new pharmacological approaches.

As basic scientist involved in research focused on a human disease, I believe that one of the most important goal of a patient European association like OIFE will be to strengthen contacts and exchanges among patients, clinicians and basic scientists of different countries. I do believe that interaction and collaboration among us is fundamental to ameliorate knowledge of the disease and hopefully to ameliorate patients quality of life.

The Costs in OI

Interview with Dr. Argerie Tsimicalis - Shriners Hospital for Children, Montreal

Who are you?

I am an Assistant Professor at McGill University. I am one of a handful of nurses in Canada who has completed a PhD with a specialty in child health (less than 1%). My relationship with OI started with the invitation by Shriners Hospitals for Children®-Canada in 2013 to establish an independent program of research, as their inaugural Nurse Scientist.



Can you tell us about your project?

I am seeking to render visible the costs associated with OI. A first step is to establish global partnerships with scientists, families, clinicians, decision makers, and OI-organisations. I am worried that families of children with OI may be bearing costs. These costs may compromise their children's best interests and impose inequitable burdens on children and families. Existing treatments, programs and policies may also be escalating these costs, severely impacting the quality of life of the entire family. Current partners are Shriners Hospitals for Children®-Canada, McGill University, the University of Cologne and the OIF and OIFE. We welcome the collaboration of other partners as well.

What kind of costs are you thinking of?

I am thinking of the various out-of-pocket expenses that families may occur because their child has OI. These out-of-pocket expenses may include accessing health services for medical, surgical, and rehabilitative treatment of their child, or paying for medications, supplies, and equipment. Access to these health services may or may not be funded by their health insurance but there are also travel expenses associated with accessing these specialized health services. While their child is hospitalized, families may need to pay for food and accommodations and to coordinate for the care of their other children; at times paying for babysitting, or need to pay for help to keep the household running.

Why global - when healthcare systems are different from country to country?

My research with families of children with cancer in Canada, India and Guatemala, has lent insight into the types of costs families of children with OI may incur. Many of the cost categories and items are the same; however, the reasons for incurring these costs, and the impact of these costs on family may differ. By using the spirit of the international OI-community, we are seeking to understand these costs across varying contextual factors such as countries, geographical boundaries, and health care systems, so we can collectively identify best practices, and together, as a global community, take action.

What is the methods used?

With Marilyn Monrad, one of my students, we have determined very little research has been done to understand families' costs. However, we do know that costs remain a real concern for families. OI-groups have been compiling stories, launching fundraising efforts, and creating resources to best support families financially. These efforts, may be captured in documents outside the published research literature, which are referred to as 'grey literature'. Examples may include: pamphlets, websites and newspaper clippings.

How will OIFE members be involved?

OI-organisations know far better than me about the costs associated with OI. I need their expertise, resources, and networks, so I can help bring this issue to the forefront. Because of this, I am reaching out to the OIFE member organizations to help identify these sources of grey literature, and send the material to me by email [argerie.tsimicalis@mcgill.ca] or postal mail [Shriners Hospital for Children®-Canada, 1003 Boulevard Decarie, Montreal, QC H4A 0A9, Canada]. If a member of the OIFE organizations would like to be more implicated, we welcome their contributions.

How can we as organisations later use the results in our policy work?

We will compile and synthesize all the literature related to costs associated with OI. Our collective efforts will lead to the creation of policy briefs to promote needed changes. However, these policy changes must be created in partnership with the organizations, so they advocate for change in their respective countries.

Do you have other projects of interest?

Oh! I have so many projects, which I would love to share with the OI-community, including our most recent "Bones and Fractures Memory Game", launch March 7, 2017 (see photo below).



The RUDY-study

Interview with Alison Turner, Project Coordinator



Can you tell us a about your project?

The RUDY Study started in 2014 and was an idea of Dr Kassim Javaid. The goals was to try to understand more about all aspects of rare disease from the patients' perspective. Some of this new understanding will be used to develop new tests and treatments. A first step is to gather information in order to describe in more detail the different types of rare diseases and what it is like to live with them. We intend to do this through questionnaires completed every 6 months. We are recruiting as many participants to this study as possible, as well as unaffected relatives. Dr. Javaid has approached many different patient groups and other clinicians to start to form a group of people who could advise and comment on the work being developed.

Who are the target groups?

The target group started out as participants with a rare disease of the bones, joints or blood vessels within the UK. We also invite children to join and complete their own paediatric questionnaires although they will need to be signed up by their parent/guardian before they can complete them. Following on from comments from our patient forum group it was decided to expand the recruitment to include unaffected relatives and family members. At the moment we can only include people from within the UK, although we are looking at the logistics of expanding this to include other countries.

How many participants do you have?

At the moment we have over 700 (ca 120 with OI). Logistically there is no limit to how many participants we can accommodate but originally we did hope for 500. We now would like 5000!

How do you use patient reported data as a method?

It's really important to focus on how the rare diseases affects a person's life. To do this we only use questionnaires that have been tested to work if patients themselves complete them. From this information, we can then describe in detail how different rare bone diseases affect patient's quality of life, sleep, pain, tiredness as well as other aspects of their life.

What kind of data are you collecting?

We are collecting questionnaires every 6 months looking at sleep, pain, fatigue and quality of life. We have separate questions for children. We also collect information on fractures, medications taken and how long it took to get a diagnosis. Currently we are building more content to include other medical procedures such as surgeries, transplants and treatments.

How are the studies financed?

The main funding for the study comes of the National Institute of Health Research Oxford Biomedical Research Centre. This is a government based funding pathway that recognises early phase research.

How are the patient groups involved?

We work with many patient groups in the UK and they have been very important in setting up the RUDY patient forum. This forum is at the heart of the study and many of the things we now include have come from our patient

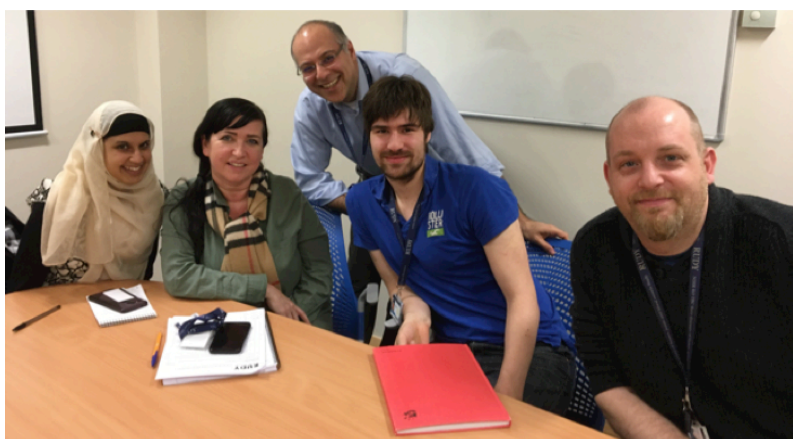
group. From the very beginning the patient forum helped design the information sheets and consent forms. We work closely with several organisations who have helped recruit for us. We also have bi-monthly skype meetings where we talk through latest ideas for the study. Over 20 people with different rare diseases (and some family members) belong to this group, but different people attend every skype. This adds to the variety of the expertise. After all the person who knows the rare disease best is the person who lives with it. All the committees within RUDY have a patient representative on them and the patient group also tests every new feature on the website. One member of our RUDY patient forum recently presented RUDY to a rare disease charity day and talked about her experience of the study: <https://youtu.be/tv5o9Kfyt08>

Why should people with OI take part in research studies like this?

The aim of RUDY is to help researchers develop new tests and treatment for rare bone diseases, including OI. Through RUDY we can measure how the rare disease changes over time, its effect on quality of life and identify different groups within each rare diagnosis. It also allows researchers from different hospitals get in touch with them with their study ideas.

Anything else you want to tell the OIFE Magazine readers?

Check out the RUDY site on rudystudy.org if you want to see if RUDY can work in your country as a person with OI, a clinician or researcher, do get in touch with us at rudy@ndorms.ox.ac.uk



From left to right:

Samiya (Data Manager)
Alison (Project Co-ordinator) Kassim
(Project Lead)
Nathanael (Project Programmer) Joe
(Project Developer)

Rare Commons

Interview with Begonya Nafria Escalera

Can you tell us a about your project?

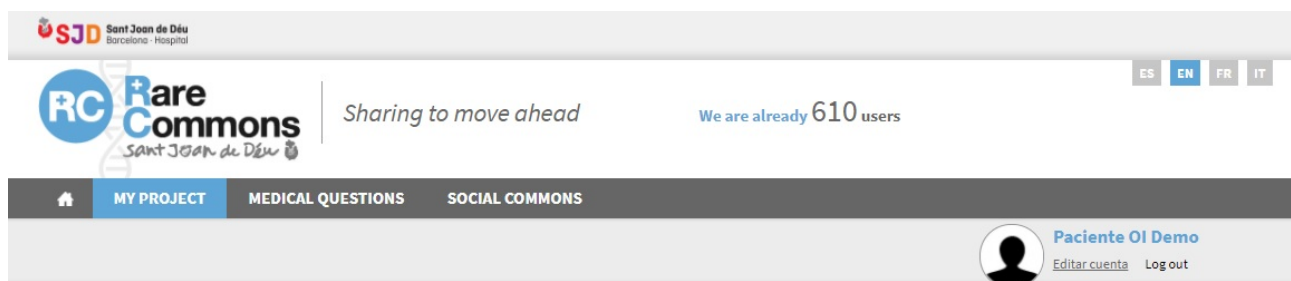
Rare Commons (RC) is a research project based on an online platform <http://www.rarecommons.org> - which allows us to obtain reliable and updated information on a particular rare disease. The different communities are based on a dynamic format modeled on virtual social networks. They are lead by an expert medical team in a secure environment and are based on information, learning and participation. Families are empowered through high quality patient oriented scientific chapters about a rare disease (in this case OI) and asked to fill in exhaustive questionnaires about their disease.



What is the content and time frame?

Rare Commons was launched in 2015 and now includes 9 different projects. The OI-project was launched in 2017 in Spanish version and includes 12 different medical chapters addressed to patients/families and the related survey for gathering of clinical information:

1. Genetics
2. The bone
3. Other orthopedic disorders
4. Orocranial alterations
5. The ear in OI
6. The eye in OI
7. Other clinical aspects
8. Medical treatments
9. Physical exercise and rehabilitation
10. Psychological aspects in OI
11. Clinical trials
12. Quality of life and PROMs




































Hi! **Paciente OI Demo**

My data

My profile	 EDIT
My doctors	 EDIT
Growth	 EDIT
Treatments	 EDIT
Analytics and reports	 EDIT
Fractures, dislocations ...	 EDIT

Osteogenesis imperfecta

	1. Genetics of OI		
	2. The bone		
	3. Other orthopedic disorders		
	4. Orocranial alterations		
	5. The ear in OI		
	6. The eye in the OI		
	7. Other clinical aspects		
	8. Medical treatments		
	9. Physical exercise and rehabilitation		
	10. Psychosocial aspects in the OI		
	11. Clinical trials		

My doctors

These chapters have been written by doctors using an easy language with the goal to empower in the self-management of the disease.

The participation in the research project takes approximately 8-9 months. Every 3 weeks we launch a new chapter followed by a survey. In this way, we provide specific education about OI so the questionnaires can be answered more accurately and we obtain more reliable information. The participants can also suggest new questions or ideas.

Who are the target groups?

Rare Commons has more than 600 users from different diseases. More than 100 patients are involved in the Spanish OI-community in RC, and the number is growing. The study is addressed to any person affected by OI, of any age group, children and adults included. We have started with Spanish but it is currently being translated to English and with funds it can also be translated to other languages.

At the end of March 2018 we expect the launch of the English community, that will allow a bigger sample of patients. If you are interested to be a member of the Spanish or English community you can fill in this form: <http://bit.ly/2FEB1Mf>

When the English community is activated, the team of Sant Joan de Déu Hospital in Spain will contact you.

What kind of data are you collecting?

We collect clinical data (of all different clinical manifestations), information about laboratory tests, radiology, treatments and also psychological and social issues. This information is analysed by a specific team of experts in biostatistics. The collection of clinical data from one patient takes their phenotype into consideration and the study of all the data from all the patients will help to increase knowledge on the natural history of the disease. The biostatistics approach to study the data donated from the patients, will allow us to have an exhaustive database that can be used to answer new research hypothesis or help in the recruitment process of patients to be involved in clinical trials.

How is the project financed?

Rare Commons won two awards that allowed the design of the platform. Later, when we created the OI community of patients, a donation from AHUCE Foundation allowed the translation of the medical information addressed to patients and families with OI. We want to thank the continuous support from this Spanish organization.

How are the patient groups involved?

We have collaboration with OI patient organizations in Spain. They have helped us with the diffusion of the project to their members and also to other organizations in Europe and South American countries.

Why should people with OI take part in research studies like this?

By taking part in research studies like this, people with OI will not only get accurate information about their disease but they will also collaborate with investigators providing their valuable personal experience. This is especially important when we want to study rare diseases, in which the recruitment of a representative cohort of patients is a challenge.



Anything else you want to add?

For us is very important to emphasize the message from the Rare Commons platform: Share to go ahead! The only way to increase scientific knowledge about rare diseases is involving patients and families as researchers. You are experts in the management of the disease, 7 days – 24 hours. And with your help we can discover reliable information that can benefit the whole community. We are waiting for you in Rare Commons! For further information please contact: rarecommons@sjdhospitalbarcelona.org

Examples from OIFE Members: AHUCE Foundation in Spain

Interview with Julia Piniella, president AHUCE Foundation

The AHUCE Foundation emerged in 2011 from the Ahuce Association and has the purpose of improving the quality of life of patients with Osteogenesis Imperfecta by promoting multidisciplinary research and by actively working in international contexts. The foundation works in areas that the association doesn't cover such as:

- research in all areas of OI
- international relations
- organization of info-workshops and seminars, for professionals and patients
- In the area of psychology, we manage clinical consultations (clinical psychology) and all the requests from patients or professionals coming from outside Spain.
- educational programs to increase employability of people with OI.



Although both organizations work together, they are different entities. The Ahuce association is located in Madrid, while the AHUCE Foundation is in Valencia. Both organization statuses are different and their accounts are managed separately.

Regarding research we're involved in - some projects are initiated by AHUCE Foundation and are supported by our scientific committee. We also finance other external projects. Researchers who are interested in receiving support get in touch send us a written proposal that should include the project's aims and its financial requirements. We then look for financing to support these projects. Any research team inside and outside Spain can contact us.

Some recent research projects that we have financed include:

- *The RareCommons Project:* <https://www.rarecommons.org/es>
- *Stem cell therapy for infants with osteogenesis imperfecta:* <http://bit.ly/2IsD47Z>
- *Impact of craneal and oral implications of OI in the actual diagnostic of the condition, and therapeutic approach,* together with the Universidad Complutense de Madrid
- *Genetic studies of 100 patients with a clinical diagnostic of osteogenesis imperfecta.*

Together these projects had a total economic investment of more than 45.000 euros.





We do different kind of fundraising events. By the end of August, four non-professional and altruistic swimmers will try to cross the North Channel. The purpose is to help us gain visibility and get funds to OI-research. One of these swimmers has OI. They will start in the Irish port of Donaghadee in the south of Belfast Lough and will arrive in the Port Patrick area in Scotland. Up until today only around 50 people have swam across the North Channel, which is

much colder (approximately 13 or 14 °C in August) than the English Channel. In the summer it is packed with giant jellyfish of the lion’s mane species that sting and can cause much pain. We have made a *crowdfunding* page for whoever wants to support the cause: <http://bit.ly/2FRoM2d>

You asked if we had other information to share with the OIFE Magazine readers. While the AHUCE association is a national association and is aimed at improving the lives of people with OI that live in Spain, the AHUCE Foundation is international and it supports people from both Spain and other countries, specially from the Latin American region and other Spanish-speaking regions. In 2017 our foundation covered over 100 international requests from different Spanish-speaking countries.

We have created an online library on OI in Spanish, with material about OI in both written form (brochures, posters, bulletins) and in audiovisual form (videos), aimed at informing and spreading knowledge about the condition. The material has been reviewed by our specialists and our scientific committee.

The Foundation also collaborates directly with teams and doctors from other Spanish-speaking countries. Our main purpose is to serve as a direct link between European OI associations and professionals and people affected by OI in Spanish-speaking Latin American countries.

MANEJO BÁSICO DE BEBÉS 1 CON OSTEOGÉNESIS IMPERFECTA

ZONAS MÁS SEGURAS
LAS PARTES PROXIMALES DEL CUERPO

ZONAS MÁS SENSIBLES
LOS HUESOS LARGOS DE BRAZOS Y PIERNAS

CABEZA
TRONCO
ABDOMEN
CINTURA PÉLVICA

HÚMERO
CÚBITO
RADIO
FÉMUR
TIBIA
PERONÉ

1 MANOS AMPLIAS
Abre siempre mucho las manos para abarcar zonas más amplias y repartir mejor la carga por toda la superficie.

2 FOCOS DE PRESIÓN
Evita los focos de presión directa sobre bordes óseos superficiales como las costillas.

3 BRAZOS DE PALANCA CORTOS
Siempre que necesite mover brazos o piernas, utiliza brazos de palanca cortos. Evita las tomas lejanas a la articulación que vayáis a mover.

4 NO FORZAR LAS ARTICULACIONES
Evitar forzar las articulaciones o sobrepasar su ángulo habitual de movilidad. No es conveniente realizar movimientos de torsión de sus miembros.

5 EVITAR MOVIMIENTOS DE TRACCIÓN
Los movimientos de tracción están contraindicados pueden ser lesivos debido a la poca estabilidad articular y a la hipertrofia propia de la OI.

6 NO MOVILIZAR DESDE BRAZOS O PIERNAS

Estas recomendaciones están basadas en la experiencia clínica y respaldadas por consenso de médicos ortopédicos y médicos que trabajan con pacientes de osteogénesis imperfecta. Se recomienda que sean tenidas en cuenta para el mejor manejo de los afectados y la disminución del riesgo de fracturas en la primera infancia. Siempre consultar los protocolos del médico a tratamiento responsable, que trabajará de acuerdo a cada paciente y con cada paciente. Cada paciente con osteogénesis imperfecta tiene unas características propias, lo mejor presentarse en siempre cuando lo mejor posible cada caso a un equipo gestor por el profesional adecuado.

AHUCE

Research without borders – an update from Care4BrittleBones

by Dagmar Mekking, Executive director C4BB

An interesting fact about research is that (after an average of 17 years) only 14% of Clinical Trials will have led to widespread change in care¹. This is surprising, as good ideas would surely be adopted by any doctor keen to improve the life of their patients, right? We don't claim to know why the impact is so low, but we do see that most researchers today are still working by themselves or in small local settings. Transferring local insights into large scaled change is generally difficult when others don't feel they have been co-creating this change. This is how humans are...also outside the medical field.

Care4BrittleBones was set up in 2012 to support research for OI, which makes a widespread impact. It should be possible, because research is without borders. New insights from research can lead to benefits for everyone with OI potentially. Therefore, **everyone worldwide is invited to engage with us who is interested in OI Research**. At the moment 6 patient organizations are working closely together with us to select the right research for funding and discuss OI research strategy, raise funds for OI research and advocate for/inform about OI research:

- DOIG (Germany)
- SVOI (Switzerland)
- ASITOI (Italy)
- VOI (The Netherlands)
- AOI (France)
- ZOI (Belgium)

A good example of “research without borders” is the Project “Physical rehabilitation for children with OI”. This 3-year project was started end 2016 by Dr Oliver Semler and Brigitte Mueller. They initiated a project to develop a Consensus paper on Physical

rehabilitation for children. The challenge: To do it in such a way that the very best expertise

worldwide was brought together. They invited the 14 most experienced experts for OI in children worldwide to work with them. They all accepted the invite! The finalized scientific paper will be published in the coming months. The project now proceeds to the next stage, where the insights gained will be translated into “practical formats” like educational and motivational videos and instructions for exercises. A web platform will be established to make all material easily available for everyone: Physiotherapists, Occupational therapists and people with OI. The material will be for free and available worldwide. More activities are planned for 2019 to keep the momentum and eventually use the experience to update the Consensus Paper in 2020 working with the group of experts which continues to support this work.

At Care4BrittleBones we love these kind of projects. We are certain that this research will be one of the 14% which will have a clear wide spread impact: It is co-created with the best experts available globally, who are all long term partners of the OI-community. Two OI-Community representatives were part of the project and there are many people ready to promote its use globally. Currently we are in the middle of a new Proposal Round which will spend at least €350.000 on OI-research. We have received project proposals worth €3,3 Million! We will select them on the basis of our communicated criteria, which includes also “Impact beyond the local level (e.g. through knowledge transfer and international collaboration)”. Only 25% of the proposals seem to meet it. Interesting...



¹ Source: E.A. Ballas/S.A. Borren Managing Clinical knowledge for health care improvement

Marie's Youth Corner

This is SO cool! I just read about a new clinical study where they will investigate a new drug called BPS804. It might improve the strength and flexibility of the bones, which means fewer fractures. It sounds amazing! I always thought that OI would be an unchanged diagnosis.

It is a totally new study and the drug is only on an experimental basis, which means that the drug has only been tried on mice with OI. But the mice showed significant improvements and the bones became stronger. This also means that they do not know all of the possible side effects of the drug.

There are some criteria's you need to fit if you want to participate in the study. First of all, you must have broken a bone within the last 24 months. Then you need to have right gene defect and then you must be able to get a scan from a special scanner. The problem with this scanner is that if you have rods in your bones they will ruin the scan. I have these stupid (and very helpful) rods in all my bones, which means that I may not be able to participate in the study. Right now I have an appointment on the hospital next week, so I can try the scanner. I'm wishing for my rods to be too short, so they will not ruin the scan picture.



I know it's a bit silly, but I really want to participate in this study!

OIFE YOUTH EVENT

24th - 28th October 2018

REGISTRATION WILL OPEN 1ST OF MAY

WORKSHOPS - PARTY
- AARHUS CITY TRIP

PINDSTRUP CENTRET -
DENMARK

PICK UP FROM AIRPORT WILL BE ARRANGED -
AGE 16-35



If you fill the criteria and get permission to participate in the study, you still can't know for sure if you will get the drug. 75% of the participants will get the drug and 25% of the participants will get placebo, which is a drug that contains no active ingredients.

Right now they are trying to find people with OI who wants to participate in the clinical study. I really want to be one of these people! Imagine if it actually works. Don't get me wrong! I love my life as it is with OI. But the thought of becoming a bit lesser disabled sounds very appealing to me...

Disclaimer: OIFE wants to emphasize that these are the words and thoughts of Marie as a young person with OI, and not an endorsement of a particular study from the OIFE.

Artists with OI: Fabiano Lioi

Who are you and what do you do?

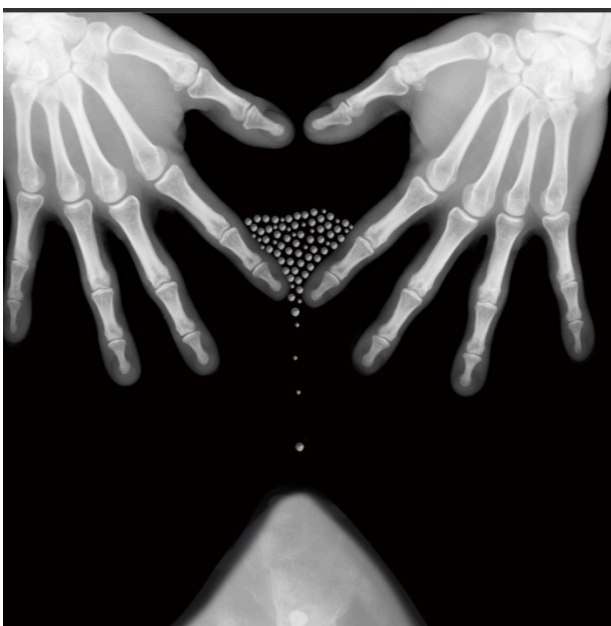
My name is Fabiano Lioi, for other people I am an actor and musician taking his first steps in the field of contemporary art. I'd rather call myself a collector of human relationships and a revolutionary. This is what I answer when people ask me what my job is...

In what way has OI affected your art?

I do not know. I have always been aware I suffered from OI and I do not know what neurotypical people (or common ones) experience in their lives. My parents have taught me to always give my best. Nothing is impossible. You just need willpower, a brass neck, a smile on your face and a little imagination. My art has been influenced by the love for life my family taught me and by my strong mood, rather than by OI.

What projects are you currently working on?

I have just finished shooting a movie, "Brutti e Cattivi", that came out in October 2017, and a TV series I have starred in for December will launch in April 2018. Whenever I am free on Sundays I go around Italy and Europe (when possible) with a performance titled "*Coloring to Hide - Unveiling to Communicate*", which you can follow on Facebook page "UAVE" <http://bit.ly/2FTb6ni>



In 2018 I will start an international crowdfunding aimed at financing my exhibition "O.I. ART IN A FRACTURE", about Osteogenesis Imperfecta through words and images.



What is "O.I. ART IN A FRACTURE" about?

"O.I. Art in a Fracture" is an art project or a contemporary art exhibition made of 20 works (scan images), two sculptures in wood, paper, glass and mirrors. There are also eleven stories (some are autobiographical) and twelve interviews with doctors, relatives and friends of people affected by OI. None of the people that were interviewed is my relative or friend.

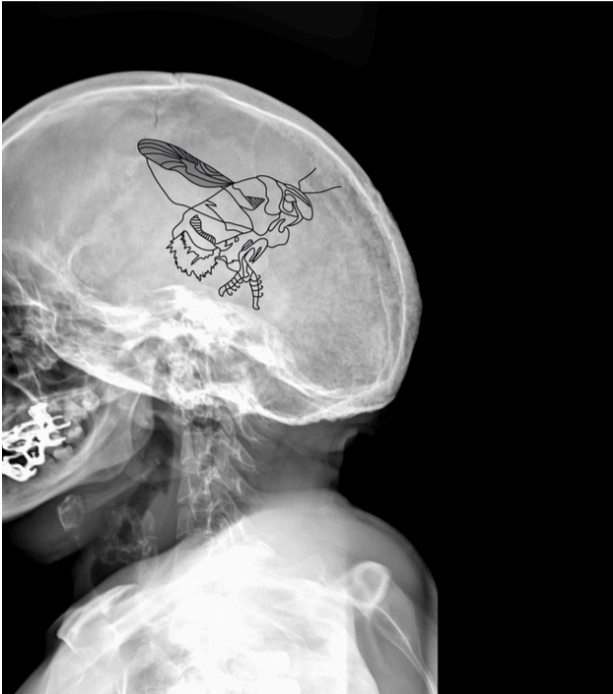
How did it develop?

One day I was looking at old X-ray scans of old fractures. I chose one and I took a picture of it just for fun, then I doubled the same image and I started overlaying several images, pointing a light at them and creating new images. I wanted to give a name to each image, so I invented a character called Kim and started using stories in order to explain the images. That's how I had the idea of depicting OI through an art exhibition.



What are your hopes for your project?

I would like to create itinerant exhibitions that travel the world starting from Rome, explaining and telling people that you can and must live a full life with OI. You can find this work in a box set and a book in English, Italian and Spanish. I will keep translating this work into the languages of all the nations that will host the exhibition.



What kind of work do you most enjoy doing?

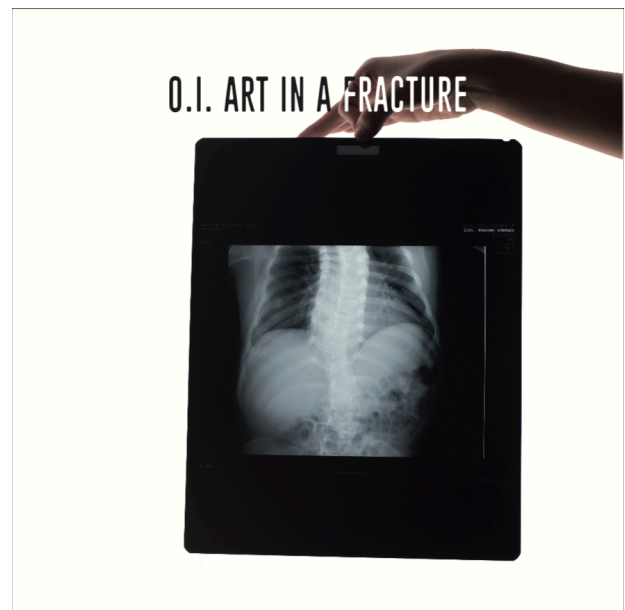
I am versatile; I like doing everything, but with one common factor: FUN. I need to have fun whenever I do something. I just love being tested.

What's your favourite art work?

All my works are my favourite ones, from the beginning till the end! Once I finish them, they are no longer my favourite ones, as they are no longer mine but belong to everyone. I am truly convinced that art does not belong to those who make it, but to people who use it and make it theirs.

What role does the artist have in society?

Artists have always been society reviewers, both in a positive and negative way. This is a wakeup call. Just think about "The Garden of Earthly Delights" by Hieronymus Bosch and the way it gives a preview of human destiny! In his first paintings Caravaggio used characters with few or none holy features and turned them into saints and madonnas. That was his personal way to criticize the Church's work at his time. The latest, real contemporary artist was John Lennon. Modern artist is no longer a reviewer of society as he is afraid of losing his fame, leaving room to mediocrity.



Greetings from India

By Laurette Paravano, OIFE delegate from France
(featured on photo page 1)

"I'm sitting in a waiting room, in a railway station in the middle of India. Our train is late and we may spend all night here..."

Fortunately, I downloaded the OIFE magazine and I have just read it. It was so 'nourishing' and inspiring!

I have been travelling in India with my husband for more than two months now, and will stay two more months in this incredible country. I should say, to quote Ingunn's last editorial: We *dare* to stay in India! Being a white OI woman is a very special experience here. Even if it is not my first time in India, it is still challenging. The rough rickshaws, crowded streets, inaccessible places are not the main reasons.



The view on different people is the point. It is very different as to what we are used to in western countries. While in western countries, people stare at me 'discretely', in India they stare 'honestly'! They express without any reservations their surprise, curiosity, or any other reaction when they see me. Yesterday, we were visiting a temple with an Indian guide, and a group of 20 or 30 people formed around us all staring at me. Some people take pictures, others ask for selfies (but this is with all white people). Children come running when they see me, very excited, some of them laughing. I can see that some people are flabbergasted and amazed, they stare wide-eyed and lose their breath. Those are not impolite reactions here. To be honest I don't know why people are so surprised. Is it because I don't fit with the stereotype of western people or with the stereotypes of disabled people?

As you can imagine, I'm used to being looked at, but not in such an intense way! Of course, I cannot change people's reactions and expect them to behave in a different way. The only one I can change is myself! I have been deeply fed up with feeling uncomfortable and spoiling my time because of this. And I realized that the only way to end it was to change my own reactions. It's hard to explain how sudden and strong it was. But it's the first time I feel something in me was fed up enough to switch. I strongly decided that I wouldn't be affected anymore, regardless the reactions of people, their number, their age or nationality. Let's see if it is a protective illusion or a real change...

It can seem obvious and simple, but it took me years to really be aware of the mechanisms that occurred in me while people were staring at me, to face them and change them. I wish I had done it earlier. But the important thing is having done it. Let's see how life tastes now!"



Announcement from the Brittle Bone Society, UK: We are turning 50 in 2018!

We are delighted to announce our **2018 conference** to mark our 50th Anniversary will be in Dundee at the fantastic Apex Hotel at City Quay on a beautiful riverside location, right beside the train station and minutes from the airport.

From **17th - 20th August 2018** we will host a series of events: a Scientific Symposium; the History Bones exhibition; talks; consultation meetings with delegates; and some serious work with our PPI groups about our OI Adult campaign and patient involvement in research.

There will also be old favourites like our gala awards celebration, and this not to be missed weekend will close with the OIFE AGM on Sunday and Monday.

Registration is now open – this includes registration for OIFE AGM 2018. OIFE delegates must contact secretary@oife.org to get a password for registration.

Accommodation and conference registration bookings must be placed separately. Full details, including registration, can be found on the conference website: <http://bit.ly/2ASA68H>

Research Announcements



Mereo BioPharma is sponsoring a multicenter international clinical trial to study the effects of the anti-sclerostin therapy drug setrusumab on bone in OI.

The study is now open to recruiting patients at multiple sites in United Kingdom, France, Denmark, Canada and the United States. Adults between 18 and 75 years old who have a diagnosis of OI Types I, III, or IV are eligible for this 1 year clinical research study, which aims to enroll up to 140 patients.

The most important inclusion criteria are:

- Genetic confirmation of COL1A1 / COL1A2 defect (can be obtained in the study)
- One or more bone fracture in the past 24 months

Visit [theclinicaltrials.gov](http://clinicaltrials.gov) listing Clinical Trials Website <http://bit.ly/2F3inxO> or the company's Study Website www.asteroidstudy.com to learn more, find your nearest study location, and register your interest in participation.

If you have any additional questions, please email AsteroidStudy@mereobiopharma.com



Research announcement update: Denosumab Trial

The company Amgen sponsors a multicenter 3 year clinical trial to study the safety and efficacy of the investigational drug **Denosumab** on bone mineral and fracture occurrence in **children who have OI**.

Currently the study is open to **eligible children ages 2-17** who have a diagnosis of OI Type I through IV.

The study has 38 sites participating in this clinical research study in the following countries: Australia, Belgium, Bulgaria, Canada, Czech Republic, France, Germany, Hungary, Italy, Poland, Spain, United Kingdom and, United States.

For more information and a complete list of study locations visit the Clinical Trials website <http://bit.ly/2kLfg3l>

If you have any additional questions, please contact the **Amgen Call Center at 001 (866) 572-6436**. Their Customer Service Representative will be able to find a site near you and provide you with the Site Study Contacts; who will tell you more about the clinical research Study.

News in brief

NEW OIFE MEMBER

Two weeks ago OIFE got our first member organization from the African continent - Osteogenesis Imperfecta SA (previously called Brittle Bones South Africa). We look forward to getting to know you better!

NEW SWEDISH OI-GROUP

We congratulate Sweden with finally establishing an OI-organization for all age groups - The Swedish Osteogenesis Imperfecta Society (SFOI). Best of luck - we're looking forward to working together. You can find them here:

<https://www.facebook.com/SFOI-1973346332695313/>

NEW OI-ORGANISATION IN BELARUS

We congratulate the people in Belarus with their newly established OI-organisation! It was formally registered in February 2018. As you can see from the photos, their first get together for children included some pizza baking.

You can find the Belarus organisation here:

<https://www.facebook.com/groups/892778114138857/>

EURORDIS PHOTO CONTEST

We congratulate Philipp & Moritz & the photographer Conny Wenk from Germany with the 2nd place in EURORDIS photo contest 2018.



PAIN IN ADULTS WITH OI

The Nursing Research Team at the Shriners Hospitals for Children - Montreal published a paper in January titled "Pain Experiences of Adults with Osteogenesis Imperfecta:

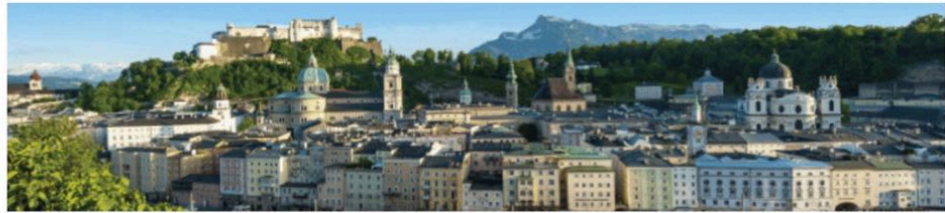


An Integrative Review.". Findings revealed that OI pain is present, problematic, and persists into adulthood.

The majority of adults experience chronic pain despite surgical, pharmacological, or non pharmacological interventions. OI pain in adults is primarily located in the back area and may be triggered from previous fractures and structural deformities. The researchers concluded that pain is a long-term symptom of OI requiring further research to better understand and manage pain in adults with OI. The article is open access and you can read it here: <http://bit.ly/2BKvHrG>

ICCBH 2019

The 9th International Conference on Children's Bone Health will be held 22-25 June 2019 in Salzburg, Austria.



Calendar

2018

April 13th - 15th	Annual Meeting DFOI, Copenhagen, Denmark
April 27th - 29th	Annual Meeting NFOI, Oslo, Norway
May 6th	Wishbone Day (International Day of OI)
May 10-12	EURORDIS AGM & ECRD conference, Vienna, Austria
May 30 th -June 3 rd	Annual Meeting DOIG Duderstadt, Germany
July 13 th – 15 th	OIF National Conference, Baltimore, US
August 17th	BBS - Scientific Symposium, Dundee Scotland
August 18th - 19th	BBS 50th anniversary, Dundee, Scotland
August 19th - 20th	OIFE AGM, Dundee, Scotland
Aug. 31 - Sep 1st	Family Meeting OI Austria, Tirol
September	Multidisciplinary Expert Forum (China Dolls), Beijing, China
October 6th	Jubileumsviering VOI (35 years), Utrecht, The Netherlands
Oct. 25th - 28th	OIFE Youth week-end, Aarhus, Denmark

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