



# OIFE Newsletter

## Issue 34 - October 2017

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### Editorial

*By Ingunn Westerheim, OIFE President*

#### **Nothing About Us Without Us**

It's soon 25 years since a group of engaged people from European OI-organisations gathered in Northampton, UK to establish a European Federation for Osteogenesis Imperfecta. Rob van Welzenis, who sadly passed away some weeks ago, had noticed that OI-organisations in Europe were struggling with the same challenges. Some of the organisations had existed for more than 20 years already. But they were not exchanging ideas about how to solve the common challenges. OIFE's honorary president wanted to change this. Rob was a visionary and his wish was that the European OI-community should work together - creating a stronger voice to get better services and treatment for people with OI. And he succeeded. OIFE was

established in 1993 and has developed ever since. Today OIFE consists of 18 European OI-organisations and 5 supporting members. In addition we have a formal collaboration with 7 organisations outside Europe, including USA and Australia. In addition we're communicating with numerous groups and engaged individuals around the globe. We will be forever grateful for Rob's initiative that brought us together in a network to share ideas and solve problems together. Thank you so much for being a visionary! You will always be remembered as the person who initiated the international community of OI.

Nothing about us, without us. It's the slogan of the disability movement and can also be connected to the constantly growing phenomenon of patient involvement. Patient participation in research was the topic of the pre-conference seminar at the 13th International Conference on OI (OIOslo2017). Same topic was also addressed in the talk OIFE and the OIF held together at the main conference, addressing more than 225 experts working with OI. The OIFE and OIF might have different perspectives on patient involvement, being two quite different organisations. But we do share the view that the organisations should be involved in research and development – on one level or another.

Why? The short answer is – because we think it makes the research and services better.

You're only doing basic research, you say? We are still interested to know about your project, the same way we are interested in participating in scientific conferences. By being informed and involved, we see synergy effects and we might be able to put you in touch with fellow researchers in other countries. Did you know that OIFE has a database of more than 900 professionals working with OI worldwide?

Other than networking– what role does the patient organisations play in research and development? How should patients be involved and why? And who are "the patients" anyway? Are they people with OI in hospital? Are they members of OI-organisations? Are they formally appointed patient representatives in reference groups or steering committees? Are they parents or only people with OI themselves? Can employees in an OI-organisation be patient representatives?



Of course they can! All of the above are examples of people with a potential of being involved in research and development. It is however important that we separate between patient participation/involvement on an individual level, on a project/service level and on a political level. Unless we are clear on which level and roles we are talking about, we might misunderstand each other and disagree without even realizing.

We all have our different roles in patient involvement in research and development. Both individuals and organisations have different resources and we acknowledge that we are not always right. But we appreciate being heard - and informed. Because...nothing about us without us. And to return to the start of it all – next August we will celebrate that it's 25 years since we came together in a European OI-community. We're looking forward to celebrating together with the Brittle Bone Society who's celebrating 50 years. We promise not to steal your glory, but we are looking forward to being part of it.

## Obituary for Rob van Welzenis - the founder of OIFE

*By Ute Wallentin, OIFE President 2002-2015 (now social network coordinator)*

Rob van Welzenis has passed away. We hardly knew about all his professional skills and expertise. For us he was mainly Rob - OIFE's president and then honorary president. The ambitious and determined father of a son with OI, who worked so hard for and with us to achieve better conditions for families with OI children...

Rob was a busy, hard working young man with enough success in his school education, studies and then in his working career as professor of physics, even before Lidy, his dear wife, gave birth to their son Taco in October 1969.

This important date later got the beginning of the OIFE – in a way...



We now skip the next 20 years. We can all imagine how a Dutch family with two children, two successful, highly educated parents, had to struggle in the 1970s and -80s after the many fractures of their little boy and his diagnosis. Taco's Mom Lidy is a doctor – but I am not sure that this fact made it easier for her...

My own parents went through a very similar, desperate situation far away in Germany – but they felt as alone as Lidy and Rob were then...

At the end of the 1980s, Rob and Lidy started to change their and our lives forever. Rob initiated the OIFE with friends in the early 1990s. He created its basic structure, the first statutes and took care of all necessary preparations that lead to OIFE's foundation in September 1993 in Northampton/UK.

He lead the OIFE as the first OIFE president through it's early years – with 7 national members (1993 – 2001); increased the membership and the importance and impact of the Federation to an international level; created the first OIFE website and functioned as webmaster until his retirement in 2016. He never failed to accompany OIFE's development and to actively support the OIFE Board as Honorary President until very recently.

The OIFE does today have 30 member organisations worldwide. We reach out to thousands of OI people, their families and friends on ALL continents and have a strong network of over 1000 professionals (medical doctors, researchers, physiotherapists and others) who cooperate with us and more and more often with each other for the benefit of ALL.

OI is still a rare disorder, but today (almost) nobody has to feel, to live and struggle with the special challenges of OI alone for very long. We can be found easily through the Internet. And the huge and international OI-community feels and acts like a big FAMILY.

It helps and supports and is there for everybody who needs it! And all this started with what Rob & Lidy and their few friends started 24 years ago in Eindhoven!

Rob, we all thank you from the bottom of our hearts for bringing all of us together and for changing the world and the lives of so many OI children and adults all over Europe and meanwhile worldwide!

You will never be forgotten and you are missed already by so many. I am speaking for all OIFE members plus their delegates, for many people from the huge OI community who met you and will always remember you.

Thank you!

## **What is the OIFE doing?**

*By Ingunn Westerheim*

**Autumn is here and for the first time in a long while, I'm enjoying a quiet week at home without a packed calendar. My mind and living room table has most of the year been occupied with my role as the Conference Coordinator of the 13th International Conference on OI, but now it is time to catch up. Below you can read about what the OIFE has been working on since the last newsletter.**

### **OIOslo2017**

The OI-event of 2017 was naturally the scientific conference on OI, which takes place every third year. The last week-end of August OIOslo2017 brought together 250 participants from 40 different countries, including more than 20 delegates from various OI-organisations around the world. For the first time in history - a patient organisation (NFOI) was in charge of the logistical part of the conference. Taco van Welzenis represented OIFE at the conference, since Ingunn was mostly busy with organizing activities as the Conference Coordinator.

Before the conference started, the Norwegian OI-organisation hosted a pre-conference seminar for representatives from ca 18 different OI-organisations. The seminar had 37 participants, with the topic of patient participation in research. Professor David Sillence and Laura Tosi were the main speakers. Various research projects with patient reported data were presented including Mereo's Asteroid-study, Rare Commons from Barcelona and the RUDY-study. We also had talks from European Patient's Academy (EUPATI) and other contributors including Lidiia Zhytnik, who's project you can read about another place in the newsletter.

Professor Sillence, who came all the way from Australia, gave a talk about how the patient organisations can adapt their information material in order to reduce confusion about the different OI-types and classification systems.





At the end of the seminar dr. Luca Sangiorgi gave an ad hoc update on the European Reference Network for rare bone diseases (BOND) and how the patient organisations can and should be involved in the future developments. He also stressed that BOND is still an infant and that expectations should be low in the first years of the network. The first phase will be about discovering the challenges in getting an OI-diagnose in Europe. Second phase includes developing guidelines for treatment and care regarding OI.

In parallel with the pre-conference seminar, Care 4 Brittle Bones hosted a Consensus Meeting for invited physiotherapists with the goal of producing a Consensus Paper on Physiotherapy for Children. They expect this work to be finalized by the end of 2017.



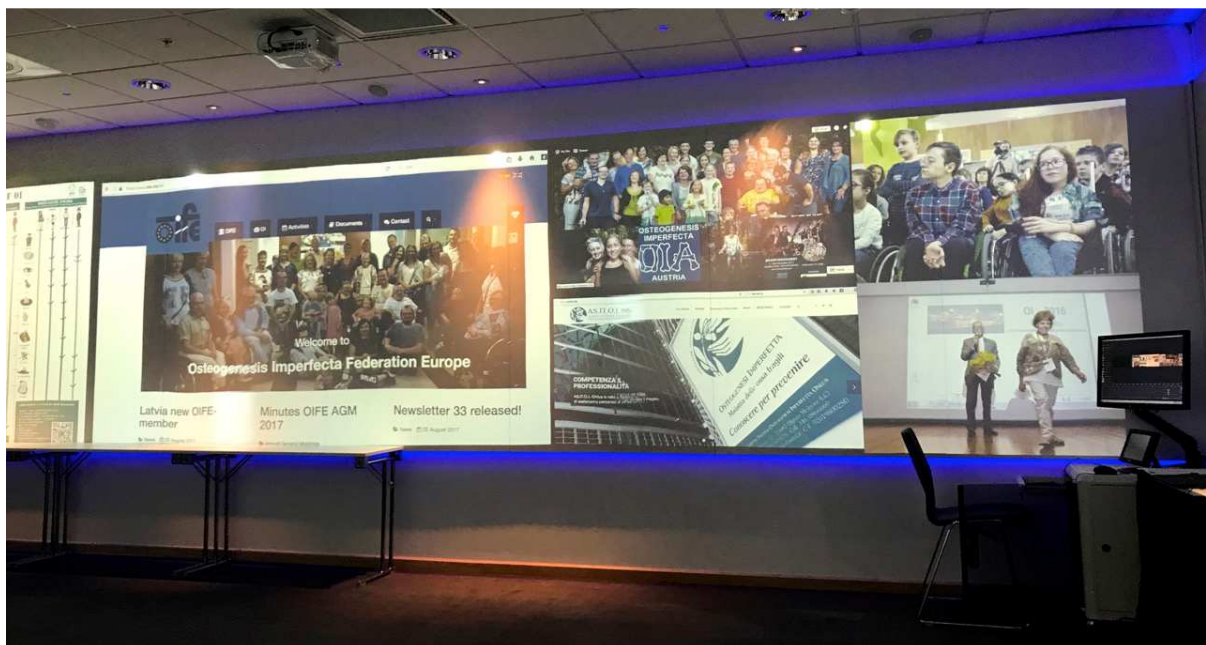
The Consensus Meeting was followed by a physiotherapy seminar open for everyone, hosted by Oslo University Hospital.

The scientific conference itself started Sunday evening and lasted until Wednesday. A lot of new research was presented - both from basic research but also more clinical. In total there were 10 different sessions with invited speakers as well as ca 35 shorter presentations from

both experienced and up and coming professionals. The full programme can be downloaded here: [www.oioslo2017.org](http://www.oioslo2017.org).

Please contact the OIFE if you are interested in a PDF copy of the abstract book! Since the target group was OI-experts from around the world, the programme was mainly consisting of contributions from professionals. But the organisations also played a small role. Care 4 Brittle Bones presented their work & goals in a morning session on Aug 29th. And OIFE's president and OIF's CEO Tracy Hart had 20 minutes to inform the participants about the organisations and address the topic of how the patient organisations can be more involved in research and development in various ways.

During the whole conference the OI-organisations were invited to present their organisations in the Inspiration Room, where photos and videos from the OI-societies worldwide were projected on the walls. Brochures from China Dolls and AHUCE Foundation were also on display as well as the Adult Health Kit from the OI Foundation.



In addition to the scientific talks, almost 100 scientific posters were presented at the conference and there were also some time for networking. OIFE's vice president Taco, was working intensely to establish a new Medical Advisory Board for OIFE. And the results from this, you can read about in the next edition of the OIFE newsletter.

### **Other meetings and events**

OIOslo2017 has had the main focus lately. In addition to 2 EC-meetings, there have also been meetings held with the BBS, Care4BB, OIF, youth coordinators etc.

- Pre-conf. seminar patient participation in research, Aug 26-27 (TW & IW)
- Pre-conference seminar on physiotherapy, Aug 27th (TW)
- 13th International Conference on OI (IW & TW + several OIFE delegates)
- Meeting with Mereo Biopharma Aug 30 (IW & TW)

## **Collaboration between OIFE and BOND**

During OIOslo2017 the European Reference Network for rare bone diseases (BOND) settled the collaboration with OIFE for the conduction of a survey on diagnosis of OI in Europe. Health care providers (HCP) in the countries who are directly involved in BOND will receive a "Questionnaire on Diagnosis of Osteogenesis Imperfecta" - one part for patients and one for specialists. OIFE has agreed to support BOND in the process of data collection, by disseminating the questionnaires to our member organisations (in addition) as well as encouraging HCP in the relevant countries to contribute with their knowledge. The results of the survey will feed BOND's "White Book" on Rare Bone Diseases 2017, which will be presented at the European Parliament 27th of February 2018.

## **OI-organisations being started in Sweden, Belarus and Chile**

We have received information that there are initiatives to start up new OI-organisations in Sweden, Belarus and Chile.

### Sweden

Inaugural meeting will take place March 17, 2018 at 13.00 in the office of RBU, St Göransgatan 82A in Stockholm. Contact person is Lars Göran Wadén–  
[lars-goran.waden@comhem.se](mailto:lars-goran.waden@comhem.se).

### Belarus

Оксана (Oksana) Симащ is trying to gather people in Belarus, who might be interested in starting an OI-organisation. Her e-mail is [Oxic@tut.by](mailto:Oxic@tut.by).

### Chile

The inaugural meeting of Fundacion OI Chile took place in Santiago de Chile on September 30th 2017. Newly elected president is Andrea Medina who can be contacted on [andreamedina@gmail.com](mailto:andreamedina@gmail.com).

**Questions?** Please e-mail [president@oife.org](mailto:president@oife.org)

## **OIOslo2017 - a report from Australia's OIFE-delegate**

*By Julie Haraksin*

After spending 6 weeks cruising the Mediterranean and the Norwegian coast, my husband Ian and I arrived at the Oslo Train Station from Bergen and headed to the Quality Expo in suburban Fornebu. We were looking forward to staying in one place for a time seeing Oslo and learning what is new in the international OI Community and the relevant scientific developments. What we delighted in was the warmth and passion of those representing the OI Community and the breadth, diversity and commitment of the medical professionals in attendance!

After a couple of days of sightseeing in Oslo the Pre-Conference Patient Conference commenced. It was enjoyable to meet the various OI representatives from many parts of the world including Europe, China, Japan, USA, and Australia.



An important focus of the Pre-Conference presenters, and indeed the dialogue that ensued, was on 'patient reported outcomes'. One such discussion noted how training by the EUPATI European Patients' Academy was being given to nominated community representatives on how to best provide input into all stages of research design and implementation including determining what should actually be studied



We also heard of the research and collaborative work by several groups worldwide. The European Reference Network BOND project is focusing on improving treatments for Bone Disorders (ERN BOND). So too the Rudy Study in the UK is working to transform clinical care for patients through patient driven research. In the USA the Rare Bone Disorder Consortium (Brittle Bone Disorders Consortium) is trying to create new and better treatments for people with OI.



I see these and other programs mentioned in the proceedings, with their expressed consumer oriented values and objectives, as a positive trend. Much is happening and people with OI are moving closer to the centre of relevant research decision making. Many of the programs are also working together with people with OI on how to improve support to individuals and families clinically and psychosocially so OIers can best live full active lives.

I was particularly captivated by the discussions around the OI Foundation's Natural History Outcomes Report. Many delegates had not heard about the outcomes of this research. The report from Dr Laura Tosi was quite detailed. I was pleased to see that the Foundation has



been using the aggregated data to guide its efforts to identify what information the OI Community and our medical practitioners need. The Foundation's response has been to develop a series of short videos on many topics from how to manage a fracture to advice for anaesthetists. These can now be viewed by anyone on the OI Foundation website.

Dr. Tosi was also keen to find out what issues and areas of research people with OI would like to see pursued. As most of us in attendance were adults with OI we focused on adult research needs:

- Cardiovascular issues
- Inform Drs of holistic patient issues (not just fractures or endocrinology issues) & how to encourage them to exchange information between specialities
- Audiology issues
- Healthy living advice – tools
- Multidisciplinary research
- Dentinogenesis imperfecta
- Women's issues
- Pulmonary issues including sleep apnoea
- Basilar Impression



Before we knew it the Pre-Conference was over and the Scientific Conference was set to commence. While there is no space here to repeat what is fully and accurately stated in the conference abstracts, I thought I'd highlight a few points that attracted my attention:

- Genetic testing which used to take 4-6 months - can now be completed in usually 4-6 weeks
- For Pre-implantation Genetic Diagnosis – clinics are now using 4-8 genetic samples from the OI parents and getting better analysis.
- The researchers still don't know why many of us are shorter than average
- Surgeons are now exploring how to treat non-union fractures, which are experienced by many adults with OI.

I also picked up some specific take home messages, which we can all apply to our daily lives:

- EXERCISE! Just MOVE! We've all heard this message so often. But there is now solid biochemical evidence that it is good for our bone density as well as our overall health.
- Make deep breathing a regular habit – because pulmonary issues are the highest cause death in OI.
- Take colds seriously – don't ignore them! It is not just an issue of restricted lung capacity in those of us who are of small stature or have scoliosis; it may also be about the biochemical composition our lung tissue.
- Participate in every Patient Reporting study possible – the more information the researchers gather the better – a larger sample size enhances validity
- Eat blueberries – biochemically one of the best foods for bone health.

Like most OIers, we are always hopeful that there will be significant advances, which will lead to an improvement in the lives of people with OI. What we discovered is that there is a growing understanding of what is happening with OI especially at the biochemical level, but as is the way of these things, many answers often give rise to many more questions. So too, many of the longstanding questions are still to be understood and in this regard there were many spirited discussions. We note that science has still not come to terms with why OI disorders with identical genetic markers (genotype) have such great variability in the way OI manifests in the individual (phenotype).



As Australians we were delighted to note the respect and affection afforded to our Professor David Silence who has for over 40 years contributed to the OI body of scientific knowledge and has provided personal support to so many of us with OI.

The Conference ended all too quickly, as all OI Conferences do. Given my own lack of an advanced science background I, at times, felt somewhat overwhelmed by some of material's complexity but the Conference papers allowed us non-scientists to gain insights into the direction and significance of much of the research. I was impressed by the depth of conviction, dedication and compassion of many of the researchers, doctors and health practitioners. Certainly we will all be intrigued to see what the next three years will bring in Sheffield.

Ian and I are grateful to many of our fellow OIers such as Ingunn, Taco, Ute, and Inger-Margrethe for making us feel incredibly welcome. We had a wonderful time making new friends who share so many similar experiences and with those who are working to create a better future for all of us with OI and our families. So for now we will bid you all *adieu*.

## Impressions from OIOslo2017

### Ossama Essawi

#### Who are you and what is your relationship to OI?

I'm a joint PhD candidate between Ghent University, Belgium and Birzeit University, Palestine. My research topic is OI in Palestine that is mainly held to identify the OI causal mutations in order to set up a diagnostic panel for this disorder. Meeting 91 Palestinian OI patients was a critical point in recognizing the neglect they suffer and the impressive courage and strength they have in facing their situation without any aid. I hope that this work will shed light on those patients and help in improving their lives. I also believe that the famous quotation by the well known author Kahlil Gibran "Out of suffering have emerged the strongest souls; the most massive characters are seared with scars." is the best to describe this group of people and what they can become.



#### What is your opinion about the conference in Oslo?

It was a very well organized conference that successfully brought together the efforts, knowledge and experience of all people working in this field from all over the world, producing a very informative and inspiring collaboration between all participants.

#### What was the highlight for you personally?

In addition to the great knowledge I got, it was a great opportunity to network and meet with different organizations and people who were very supportive and enthusiastic to help improving the Palestinian OI reality. I am very thankful to all contributors in this field and especially to the successful collaboration between the clinicians, scientific researchers, OI organisations and patients that are supporting and sponsoring the progress in this field.

### Dr. Nadja Fratzi-Zelman

#### Who are you and what is your relationship to OI?

I'm a bone biologist at the Ludwig Boltzmann Institute of Osteology in Vienna, Austria, headed by Prof. Klaus Klaushofer. Our research group focuses on bone tissue characterization at the submicroscopical level to improve our understanding of bone fragility. Typical analyses are the evaluation of mineral content and composition of the collagenous matrix. Since bone fragility in OI arises not only from reduced bone mass but also from inherent alterations of the bone material quality, we focus on the latter in our research. For example, we discovered that hypermineralized bone matrix is a hallmark of many forms of OI, independently of mutation type and clinical severity, in humans as well as in murine bone. The increased mineral content most likely makes the bone tissue stiffer than normal and, therefore, prone to fractures.



### **What is your opinion about the conference in Oslo?**

The conference was a unique opportunity for me to meet and discuss with many specialists and learn about the latest developments in the field. In general, more specialized conferences, such as the OI Oslo 2017, are a more effective vehicle to transport ideas and knowledge than large medical conferences with many parallel sessions. In particular, the session on “The connective tissue spectrum in OI” broadened my view on OI from a skeletal disease to a syndrome with cardiovascular, pulmonary and complex dental conditions. Finally, Dr. Marini gave an impressive overview on “What is OI in 2017” delineating where we are in terms of research and where we are going.

### **What was the highlight for you personally?**

Most enriching for me as bone biologist was the opportunity to meet and interact with people with OI and learn about their feelings and ways of living with this condition. I would like to thank the organizers for inviting me and in particular Dr. Lena Lande Wekre and Ms. Ingunn Westerheim for their very warm welcome and their hospitality in Oslo.

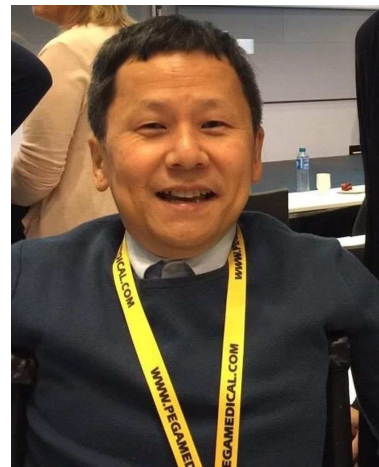
## **Toshi Seto**

### **Who are you and what is your relationship to OI?**

I am a pediatrician in Japan and also I have OI type 1.

### **What is your opinion about the conference in Oslo?**

The meeting was an exciting experience for me. Although I have OI, this was a first time to visit this kind of OI-meeting. I was so excited to see hot discussions and I have changed my idea about OI through talking to people having OI, their families and many specialists in this meeting. I think positive now. I really want to join next meeting 2020 at Sheffield and I am going to study OI from now on.



### **What was the highlight for you personally?**

I could know overview of progress of research and treatment for OI.

## **Sandra Webb**

### **What is your relationship to OI?**

I'm a Patient Services and Retention Manager for **patientprimary**. We are currently working with Mereo on OI studies. My interest was to learn more about OI with a particular interest in patient advocacy to listen to their clinical trials experiences which we can use to enhance our service providing patients travel, accommodation and reimbursement claims for patients and their carers, partners and parents.





**What is your opinion about the conference in Oslo?**

A very extensive programme covering everything from diagnosis, research, patient organisations, current status in treatments, role of clinical psychology to future optimal treatments. For a first time attender, it was a very comprehensive insight into OI, with a great list of invited speakers, oral communications and posters.

**What was the highlight for you personally?**

Meeting some incredible people with many talents who are a real inspiration in particular Dr Seto Lecturer in Department of Paediatrics in Osaka and his wife, who presented a poster on A novel COL1A1 mutation in a Japanese family with OI. Listening on what he had to do to attend the conference and how it has inspired him to think about retraining in managing OI patients in hospital.

## Research Study Announcement - Prenatal Diagnostic Testing

**Are you a parent with OI or the parent of a child with OI?**

Some parents who have Osteogenesis Imperfecta or parents who have babies with Osteogenesis Imperfecta have testing done before the baby is born. Many parents consider testing. We would like to learn more about what parents think about these tests. Parents will complete a brief survey that takes most people about 15 minutes. The information will be used to teach genetic counselors and doctors how to better work with parents.

To participate in this study, you must:

- Be a biological parent with Osteogenesis Imperfecta
- OR a biological parent of a child with Osteogenesis Imperfecta
- Read and write English
- Be 18 or older

**To participate please register before end November 2017:** <http://bit.ly/2yGHcwB>

If you have questions, please contact Rachel Sullivan at [Rachel.Sullivan@cchmc.org](mailto:Rachel.Sullivan@cchmc.org) or Kara Ayers at [Kara.Ayers@cchmc.org](mailto:Kara.Ayers@cchmc.org).

*Privacy: Your participation in completely voluntary. Nothing will happen if you decide not to take part. You can stop participating at any time. Your name will not be shared and your answers will be private.*

**Disclaimer: The OIFE is not involved in the design or management of this research, and as such, is neither endorsing nor supporting this study. One of the goals 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.**

## Science: Interview with Lidiia Zhytnik

### What is your relationship to OI?

My name is Lidiia Zhytnik. I am 28 years old and live in Estonia. My parents come from Ukraine, but I was born in a small town on the North-East of Estonia – Sillamäe. After I was born, my mother was told that I have Osteogenesis Imperfecta, as well as herself, and my older sister. Now we know about at least four generations of OI in our family. I was treated at the Clinic of Traumatology and Orthopedics of University of Tartu since my childhood, by our Estonian OI doctor Dr. Katre Maasalu.



### What is your job/profession?

I was always curious about my OI and compared myself to my sister, who had no fractures during childhood, but was told to have OI too. It seemed very mysterious to me, so I started to study biology and genetics. I studied gene technology at the University of Tartu during my Bachelor's study. Then I continued, with Master's degree in biomedicine. From time to time I had to visit my doctor. I remember, as I was asked to show my blue sclerae to medical students. And I was always happy to do so, and to help educate medical professionals about OI. Once, our family was invited to participate in the OI study, which included genetic analysis. I told Dr. Maasalu, that I study genetics and I have interest in OI. Now I am a 3<sup>rd</sup> year PhD student in the Clinic of Traumatology and Orthopedics of the University of Tartu and work with Dr. Maasalu for five years already.

### Can you tell us about your research project?

We have a database of 250 OI families from Estonia, Vietnam and Ukraine. We gathered samples of blood for DNA and RNA analysis and genealogical information and detailed phenotype descriptions. It's a very promising and unique database, which contains a lot of information for future research.

The goal of my research project is to describe genotype-phenotype correlations in OI and intrafamilial variation of OI severity. The thing, which seemed mysterious to me in my childhood about my sister's OI and my OI, actually is still mysterious for the scientists. Some of the investigators believe that differences in OI severity between family members might come from epigenetics, others think that it might be hidden in modifier genes. There are many factors, which contribute into bone strength, and all of them must be taken into consideration to understand the nature of OI. I believe that understanding of intrafamilial diversity is important. It's kind of biomimicry. If we will understand what mechanisms compensate bone fragility in affected family members with less fractures, we could use it in the future for treatment.

We are collaborating with Hue University Of Medicine and Pharmacy(Vietnam). We also collaborate with Ukrainian Association Of Crystal People and Kharkiv Sytenko Institute of Spine and Joint Pathology (Ukraine).

Also we are planning collaboration with VU University (Netherlands). I am very excited about it, because during my Master study I did an internship in VU University in the research group of connective tissue disorders with Dr. Dimitra Micha and Dr. Gerard Pals.



### **Which methods do you use?**

We started with identifications of OI collagen type I mutations in the youngest affected family member from each family in our database. Now we are going to continue with panel and exome sequencing for patients without collagen mutations. Afterwards we are planning to look at collagen I expression and methylation of collagen genes, which regulate this expression.

### **How many families are you in touch with in Estonia?**

In Estonia we have 30 OI families. We suppose that it is almost 100% of OI families, which live in Estonia now.

### **How did you get in touch with families in Vietnam and Ukraine?**

We got in touch with Vietnamese OI families via Hue University of Medicine and Pharmacy. As for Ukrainian families, I met the head of the Ukrainian Association of Crystal People – Lyuba Petrova a few years ago in social media. Then I helped them with understanding of OI genetics and translation of some research papers. I feel special connection to Ukrainian OI people, as my family comes from Ukraine and part of my relatives belong to Ukrainian OI community.

### **What is the most rewarding working on this project?**

For me the most rewarding thing were people, whom I met during this work. I feel very lucky to work with Dr. Katre Maasalu and Dr. Aare Märtson, who are my supervisors. I very much appreciate their care and support. They do a lot for my whole family and it's hard to express my gratitude with words. I am glad that I had an opportunity to work with and learn from Dr. Dimitra Micha and Dr. Gerard Pals. I never thought that people could be so warm and friendly. And at last, but not least, I have met an OI family. Special thanks to Ute Wallentin, Inger-Margrethe Stavdal Paulsen and Ingunn Westerheim for help and support.

### **What has been the biggest challenge?**

The goal of the project is really ambitious and it might take more time and resources, than I thought at the beginning. Genetic studies are expensive and for rare diseases it's a special issue. But I hope that in cooperation we will overcome all the difficulties and reach our goals.

### **What are your thoughts on patient involvement in research?**

There are pros and cons to participate in the research. Of course, the main priority has to be your health and it's up to you to decide. For me, the answer is definitely YES, because I understand, that there will be no promotion in research and treatment strategies without our participation. Only we can do it.

### **Do you have a message to the readers of OIFE newsletter?**

I am very happy, that I have met the OI community. I am very proud to be part of you. You are inspiring me. You are awesome.

## **Marie's Youth Corner**

### **"What are you looking at?"**

"Wow she has a big head", a little boy shouted when the elevator opened and I came out. Hmm... Okay I didn't think my head is that big, but it must be all the wisdom it contains. I stand out from the crowd, because what is new and different attracts attention. Sometimes my friends get really shocked, because of all the staring and at times also pointing fingers at me. Often they ask me if I find it annoying that people are staring at me. To be honest I don't think about it very much. I have always been used to people staring. If I have a bad day, I obviously find it annoying that people are staring that day, but at the bad days there is so many thing that can be annoying.



Children are undoubtedly the best to handle it. "You look small!" "OMG that is such a cool tractor she is driving!" "Why do you look so weird?" Children ask all these weird things. I do find it a bit amusing honestly, when they bump into something, just because they are staring at me and not paying attention to their surroundings. It has happened a few times...

The good thing with children is that they just say whatever that pops up in their brain, and then we can take it from there. It is so much better than the parents who pull their children away, every time the child looks at a disabled person. I know they do it with good intentions, but if a child gets pulled away every time it sees a disabled person, it is obviously that the child gets a weird relationship to disabled people. Of course there is a line for what is okay to say or do. For instance when a child runs after me in the supermarket, it would be nice is the parents would step in.

There are so many people who think that it is offensive to me, if they ask me what my disability is. It is not at all offensive to me! Just ask! The faster I can answer all these uncertain questions the faster I can tear down the wall where it is my disability that is in front.



Knowledge is the best way to break down prejudice. When you get to know me, you almost forget that I am disabled. That is where it is means a person who is in front and NOT my disability.

## Examples from OIFE Members: Camps for children with OI in Russia

*By Aleksandra Romanycheva*

The Russian OI-Association “Fragile People” (see webpage [www.osteogenez.com](http://www.osteogenez.com)), that was founded in 2014, has been realizing several projects this year such as a new rehabilitation program (in cooperation with the Cologne University Clinic) or the first EURASIA Orthopedic Forum. This report is about the two camps for children with OI that took place in 2017:

The Russian Fund «Fragile people» helps people with Osteogenesis imperfecta and other bone diseases. It was founded on October 25, 2014. The fund organizes medical care for OI-children and adults and works with government to make treatment for OI-patients more available, buys wheelchairs and crutches, carries out groups of psychological assistance for parents and makes camps for children and teenagers.



We have already organized six camps. And this summer we had our seventh, «Camp on the sea» in Sochi from 15-24 of August. We usually invite children from other Russian speaking countries. This summer camp we had a visit from Nurbolat Gabdullin from Kazakhstan and one of tutors – Andrey Pashenko from Ukraine. And in the future we have an aim to integrate both OI children and non-disabled kids in our summer camps.

In april 2017 we organised a camp for kids with OI in the Vladimir District called **“The Wizard’s cup”**. It was the biggest camp ever done in Russia for OI-children (45 children visited camp). The camp's concept was based on the universe of J.K. Rowling. Kids were involved in the mystery of the Harry Potter world and all participants were divided into 4 faculties. Every faculty had its own home, decorated by the team taking part in the camp's competition to win the main prize - Wizard’s cup. Beside this, the kids were involved in self-improvement stories. They earned special prizes for developing one of 5 types of personal magic powers - power of help, creation, communication, independence and bent.



We made the educational part of our camp with «ОбразЖизни» [The way of Life]– the winners in «Innovation in education 2014». Kids were learning about a variety of professions, such as: multiplier, neuroengineer, script-writer, watchmaster, marketing specialist, TV reporter, fashion specialist, DJ, journalist, designer and others.

Seminars were held by professionals in every sphere of life. We invited a famous Russian channel1 journalist - Irina Rossius, scriptwriter - Elena Kiseleva, Aleksandr Ross Johnson - who taught children how to concentrate and others.



Our aim was - to show that people with disabilities have a lot to accomplish in a future professional life. To present their personalities kids constructed their own web site. We also invited journalist with OI – Alyona Merkulyeva, and made a skype conference with OIFE President Ingunn Westerheim to show children - you can become anything you want.

Parents parallel program included plenty of activities. Most of them were targeted to keep the idea of children's independence. We tried to show the parents - kids may do things on their own way. Moreover - sometimes they *need* to live on their own, make self-dependent decisions, take self-care and care of their mates, have self-esteem, be brave and spontaneous. To make it real, Aleksandr Kholmogorov (sportsman with OI) spoke about motivation, Anrey Grigoriev (famous coach) taught how to make wishes come true. Two psychologists were invited to accompany parents, they helped to solve issues that appeared and increase parent's consciousness.

Despite of it, we wanted parents to feel better, rested and relaxed. That's why the program included yoga twice a day, lessons to practice the contact with the body, makeup masterclasses and emotional sharing of experiences.

In 2015-2016 years fund has raised 4 821 136 rubles (70 899 euros approximately) and has helped more than 180 people with OI. To make the "Wizard's cup" camp real we raised 2 126 075 rubles. We collaborate with TV-channel NTV and made a video report about people with OI, spoke about camp on the radios "Russia" and "Mayak", used crowdfunding platform "Planet". We also received donations from private companies and ordinary people.

We wanted to make children with OI (especially those who can't walk only because they afraid to) feel strong enough to walk, probably with the help of quad cane. But for the most part - for a first time in their life. And a lot of children begin to walk in our camps.

From the other side, we wanted to make connections between children all over the Russia, to give them opportunity to share experience, feelings and thoughts, communicate with someone like them. We do this quite successfully. All children communicate with each other after camps and wait for the next one. The biggest challenge for the kids was to be separated from parents and their usual life. In the camp, children dive into a new atmosphere, creative and full of freedom. Kids got an opportunity to show their individuality, make a professional choice, find the things that they like to do. Sometimes it's really hard to make it on your own.

### **Artists with OI: Interview with Philippe Rahmy (†October 1, 2017)**

**Someone who secretly wanted to explore the galaxies as a robot, must have been at least a humorous and profound human being on this earth. To me, it is still unbelievable that this interview that Philippe gave to the OIFE a few weeks ago, was maybe the last one in his life, that was way too short.**

**Who are you and what do you do?**

I am Philippe Rahmy and, as far as I can tell, I



devote myself to language. Words became my arms and my legs, as well as an access key to the open world; my writing offers me a second body which is able to achieve everything, without any restrictions, except the restrictions of my own mind (they are huge, of course!) my attention and dedication to language explains my poetic core. I am a poet. But I also write fiction and non-fiction, because I like to explore new worlds, to find out how they communicate.

**Does OI play a role in your books/essays?**

I don't hide that I have OI in my books, but OI has never been my main subject. I would have been a writer without OI. But the way I am receptive to human vulnerability, is certainly impacted by my own experience of pain. Let's put it this way: my style, as a writer, reflects the way I see things, and even more my own ability to embrace the whole spectrum of the human condition. It is a matter of empathy. When I can't describe something in words, I have to look better. I have to feel better. OI developed this ability of feeling. Suffering is an open door leading to empathy. This door also leads to revolt. So, here I am: empathic and revolted, as are all artists, as we all are. But I do my living from it.

**You won the Swiss literature prize this year. Can you tell us about this award?**

You are kind to ask me about that award. I got it for my book entitled "Allegra", a novel telling the tragic love story of a father. This prize is an important one in my country. I feel honoured to get such a distinction. But I never forget two things: first, even when you were born with OI, in a family where art is absolutely not a matter of discussion, in a small village, you can, with some luck, a bit of talent, some empathy, a lot of humour and even more revolt against your limitations, move forward and become the person you dreamt about. Someone by your own. You can give birth to yourself; and then, when people tell you that you achieved something, you must look through them and through yourself, to continue moving, to forget the warmth of the applauses, to return to your solitude, to your quest. To my literary quest.

**What projects are you currently working on?**

I am currently working on a book about people who were wrongly convicted and imprisoned in the USA, and who have been freed after having spent a long time in prison. I have met a significant number of them, listening to their stories, but also to their long and painful silences. When the good moment arrives between us, when some mutual trust exists, I try to discuss the matter of forgiveness with them.

**Why do you do what you do?**

There can be no "why", nor any answer to such a question. I don't need any reason to fight injustice with words. Injustice hurts me more than OI. Even as a child, I was more revolted by the sufferings from other people around me, than by mine. I write to recreate the world.

**What themes do you pursue?**

My main theme is authority. On any level and in all circles. Most of the time, in the private as well as in the public spheres, authority is related to hierarchy and power. This pattern has to be redefined. Literature offers you a way to weaken the strong and to empower the weak. This could be said in one sentence: the poor have rights, the rich have duties. Let's start from there.



### What's your favourite art work?

The one I will discover tomorrow.

### What role does the artist have in society?

We, as artists, must be convinced that we will change the world. Since ever, artists are meant to distract kings; nowadays, they are supposed to distract or to educate the people. I disagree with these two roles. We have to transform the society. Art is politic.

### What is your dream project?

To be transplanted into a robot and explore the galaxies or to become a boxer. If these projects fail, if I have to be more realistic, then I would like to buy a kayak and explore the rainy forest. If this is also too much, well... I would love to be able to write a few more books.

## News in brief

**INFOGRAPHICS:** How to explain OI in only five minutes? Infographics can be a great way to do it. Take a look at the one [Fundación Ahuce](#) has made and practise your Spanish at the same time!

### ROMANIA:

In Romania, the OI treatment for children is now fully financed by the state. Following a request for information from the Romanian OI-association, the Romanian authorities provided 33 children with OI treatment: 28 of them receiving rods, and other 13 receiving pamidronate infusions (the difference stems from the fact that 8 children received both types of treatment); We consider that OI treatment in our country is following a good path, and we hope that in the near future every child needing such a treatment can benefit from it.

### RODDING IN RUSSIA:

The Russian OI-organisation chaired by Elena Meshcheryakova has been working tirelessly for five years to have the Fassier Duval rods approved by the Russian health authorities. In September 2017 their struggle was crowned with victory.

**¿QUÉ ES LA OSTEOGÉNESIS IMPERFECTA?**

La osteogénesis imperfecta (OI) es una enfermedad congénita caracterizada por una mayor fragilidad ósea.

Es una patología poco frecuente que se estima puede afectar a 1/15.000 o 1/20.000 personas.

Como consecuencia de una mutación genética, el organismo genera menos colágeno del necesario, o de peor calidad.

Es una enfermedad crónica que actualmente no tiene cura. Aún así, existen tratamientos eficaces que mejoran la calidad de vida de los afectados.

Además de la fragilidad ósea, existen otras manifestaciones clínicas:

- Estatura baja
- Dolor crónico
- Dentinogénesis imperfecta
- Pérdida de audición y visión
- Deformidades óseas
- Escoliosis.

Actualmente, la atención a las personas con OI abarca los ámbitos siguientes:

- tratamiento farmacológico
- seguimiento ortopédico y quirúrgico
- tratamiento fisioterápico
- intervención psicológica
- tratamiento odontológico
- consejo genético.

La investigación abre nuevas vías para el tratamiento de los pacientes.

**FUNDACIÓN AHUCE**  
Osteogénesis imperfecta

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## Calendar

### 2017

October 26th-27th: Council of Federations-meetings EURORDIS, Paris, France

November 1-5: OIFE/VOI Youth Event in the Netherlands

### 2018

February 28th - Rare Disease Day (worldwide)

March 9th - 11th: Executive Committee meeting OIFE (place not settled)

April 13th - 15th: Annual Meeting DFOI, Copenhagen, Denmark

May 6th: Wishbone Day (International Day of OI)

May 10-12: EURORDIS AGM & ECRD conference, Vienna, Austria

July 13<sup>th</sup> – 15<sup>th</sup>: OIF National Conference, Baltimore, US

August 17th: BBS - Scientific Symposium, Dundee Scotland

August 18th - 20th: BBS 50th anniversary & OIFE AGM, Dundee, Scotland

Aug. 31 - Sep 1st: Family Meeting OI Austria, Tirol

October 25th - 28th: OIFE Youth week-end, Aarhus, Denmark

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