Editorial

As an OIFE Vice-President, I have the honor of writing my second editorial for the OIFE newsletter. It has been a pleasure acting in this role for quite a few years now already. We are an international community of exceptional people, and I quite enjoy knowing all of you and hearing how things are going in various European and even more far-away countries. I am in particular excited about our new president Ingunn Westerheim who has already brought her energy and competence to bear on our most pressing issues.

With such a rare disease being in question, it is important for us to collaborate on the broadest possible basis. Not only can we provide peer support and share ideas across borders; one of the most important issues of concern for us is medical research. I have taken an interest in that field in particular and have promised to be the main OIFE EC contact point for researchers and drug companies. The OIFE is large enough to build a name for itself within the community of rare disorders so that, in collaboration with other such groups, we can be in contact with relevant businesses, academia and authorities.

The most important advance in treatment of OI in recent decades has been the use of bisphosphonates, which were originally developed for the treatment of osteoporosis, a costly and common affliction in the elderly population. As OI in itself is too rare for it to be profitable for drug companies to seek treatments for it specifically, it is important for us to support research that seeks to repurpose otherwise financially viable drugs.

Another important avenue to pursue are so-called orphan drugs -- medicinal products which the pharmaceutical industry has little financial incentive to produce under normal market conditions. It would be unfortunate for a drug company to just sit on a patent without making use of it because of this, if it was useful for OI treatment. We need to be aware of these kinds of opportunities and to encourage the orphan drugs' research and use.
Finally, there are the completely new treatments that are out there on the horizon. Treatment options such as use of stem cells and new bone metabolism altering drugs are promising, but these need to be thoroughly researched due to their possible risks. It is important for us to be involved in providing patient expertise and a point of view in their development.

I can’t wait to be involved in all of this, as the future looks more promising than ever! I wish everyone an excellent start to the autumn!

Eero Nevalainen
2nd Vice-President

What is the OIFE doing?

By Ingunn Westerheim

Even if summer is usually pretty quiet, there are still some activities at the OIFE virtual office. Read more about the different activities and events below.

Meetings and events
The OIFE has attended the following meetings/conferences the last 4 months:

• Meeting at Brittle Bone Society Headquarters in Dundee, Scotland – May 25th
• EURORDIS Annual General Meeting, Edinburgh, Scotland – May 26th
• ECRD-conference (EURORDIS) in Edinburgh, Scotland – May 26th – 28th
• SVOI (SwitzerlandOI) 30th Anniversary & AGM, Campus Sursee, June 3rd -4th
• “First Friday”-meetings with OIF, BBS& Care4BB
• Skype with Care4BB – June 10th
• Phone conf. with European Medicinal Agency (EMA), July 4th & Aug 31st 2016
• Meeting International Osteoporosis Foundation (IOF), Switzerland, July 21st
• BBS Family Conference & AGM, Reading, UK, Aug 12th – 14th

OIFE is getting more involved in research
The OIFE is currently involved/getting involved in three different research projects:

• Patient representative involvement with the European Medicines Agency (BOOSTB4)
• Member of the trial steering committee in project regarding teriparatide and zolendronic acid (Scotland, UK)
• International anti sclerostin trial (BPS804)
New OIFE-database of professionals

The OIFE has created a new and more expansive contact list to professionals. The purpose of the database is to spread our general newsletter (4 times a year) as well as information about scientific events and research grants. We will not use the list for other purposes or share it with anyone outside the OIFE secretariat. Do you know a professional who should be included in the list? Tell them to contact office@oife.org.

Bisphosphonates in Ukraina

The relatively new OI-organization in Ukraina (Association of Crystal People – UACP) is working hard to improve the treatment for children with OI in their country. At the moment, use of bisphosphonates is considered illegal because the local health authorities have not approved the drug for children with OI. In all other countries except Japan and Italy (as far as we know), bisphosphonates are given “off label”. This prevents the drug from being administered to children with OI in Ukraina, who only allows “approved” drugs. We are working together with UACP to solve the situation. Do you have tips on how we can solve this? Please contact office@oife.org. See also Ukrainian webpage: http://ostimperfecta.wixsite.com/oiua.

European Reference Networks

An application to form a European Reference Network (ERN) for rare bone disorders (BOND) has been sent to the EU Commission in June 2016. The Commission will decide about approval during 2016. The initiative is lead by dr. Luca Sangiori from Bologna, Italy and includes 1 ERN with 2 different sections (skeletal dysplasias and metabolic bone disorders).

OIFE AGM

We remind you to save the date for the next OIFEAGM taking place in Lisbon from October 9th–10th (meeting on Sunday AND the entire Monday) after the 1st Hispanoluso Congress about OI.

Registration for scientific OI-congress: http://bit.ly/1YnXv9e
Registration for OIFEAGM: http://bit.ly/1Ux5mMm

Questions? Please e-mail president@oife.org
My trip – risky but with a happy ending

By Qusai Al Rifai

My story begins back in 1986 when I was born into a Syrian family in the city of Damascus. Since I came to this life I suffered from a rare medical disorder called Osteogenesis imperfecta Type III (Brittle Bone Condition).

I always wondered "why can't I play normally as the other children do outside?", "I wish to go to school and make new friends!, to run very fast and that no one is able to catch me, to dance, to fight, to jump, to go to bed or get up alone!".

While growing up, I started to dream of travelling outside my country, of having a better life and new chances. Two of my biggest dreams were to continue my studies and to have a normal social life as a healthy person.

I tried a lot in order to travel to Europe but unfortunately failure was my only ally! At the beginning of 2011 the Syrian Revolution flamed out and my life turned dramatically from worse to worst because of the civilian conflict. I realized that I had to flee from the country immediately and that there was no further hope for me of staying any longer. First station of my trip was in 2013 when I decided to cross the Syrian-Lebanese borders, then take the airplane from Lebanon airport towards the Turkish airport. I lived in Turkey for 2 years and a half at my older sister house. I tried to be in contact with the UNHCR, but they promised me a lot of things and after several phone calls they informed me that it was impossible to offer me any kind of help. I could stay in Turkey and receive 150$ per month if I wanted. I answered that my case was bigger than this amount of money! That it was a matter of life or death and I refused the offer and ended the phone call. Then I gave up all for the international organizations and the embassies and I took my fatal decision to cross the sea. It was the final solution for my misery. My sister, she is unable to provide me with any type of help, she was hopeless in front of me and I was utterly lost!

In the meanwhile and in my state of confusion I was working on the other side to find some person ready to aid me across my journey to Greece, and I promised myself that I will be the responsible for his travel costs in return for his favour.

I found the guy and my future friend Ahmed through a friend of mine who introduced us to each other. We planned that first me and Ahmed should travel towards Greece by crossing the border through to the Turkish side, but the Turkish police finished the sit in and arrested most of the people, so we dropped that idea. Later Ahmed called me to ask me, if I still wanted to cross the sea and that everything was ready regarding the smuggler to arrange the trip. I answered 'yes' without any thinking!

In 48 hours I bagged my necessary stuff and met with Ahmed at the bus station. We arrived to the gathering spot after 17 hours from Istanbul to Didim. At 7 o'clock in the evening the weather was extremely windy and cold, the darkness wrapped the place, at that time I was observing the sea waves how strongly they were hitting the shore
and I said to myself "This is my last chance, I don't have any other choice to make, if I survive tonight it is because of my charming luck and if not, I really don't care anymore". Meanwhile the smugglers started to fill the rubber boat with the refugees, they threw my wheelchair into the water, threatened us with their firearms that if someone would change his mind they would not hesitate to put a bullet in his forehead and throw his dead body into the sea!

In the beginning of the journey one of the women stumbled and fell down on my left arm inside the boat, which resulted in a fracture. After three and a half hours in the middle of the sea and with 45 passengers in the boat, and all the lower part of my body submerged into the water, having also two more fractures meanwhile in my thigh after I fell down onto the rocks while the guys were holding me, we arrived finally to the Greek shore on the other side in the evening around 22:30 o'clock. From there we continued, me and my friend, by ferry to Samos island, then again from there to Piraeus port in Athens with a large ship. Many friends of mine from all around the world that I had knew before the trip and until till now gave me huge support throughout the time of my stay in Greece for three months and a half at Eleonas refugee camp in the central part of Athens.

With the help of several new friends in Greece that I made with the help of my old friends, I entered the relocation program for resettlement into another European country. I chose the Netherlands as the first desirable option on the list and my request has been approved.

I was under the MSF doctors’ treatment and care at Eleonas camp after I had a terrible accident leading to ten more fractures all over my body, when I fell down on the ground from my wheelchair.

On 15th of February 2016 the date of my flight with Ahmed to the NL had finally come, at 3 o'clock of the night we headed to the El. Venizelos airport to take our first flight - a transit via Turkey, then the second flight directly to Schiphol airport in Amsterdam. At the same day of my arrival I obtained my five years resident permit and later on I've been transferred to an Asylum Refugee Center located in the North of the Netherlands, waiting there now for my new home.
My dream has finally come true. I now want to study, work, enjoy my freedom and my independence. I want to live my life as a productive and influential person, not only be a consumer and an unknowable figure.

Recently, the doctors in Netherlands discovered that I have a serious fracture in my neck, that happened probably during my difficult trip and the accident I had back in Greece that I had. I must be careful and wait for the doctors what they will say and decide, hopefully I can overcome this problem too!

This is a short version of my long and hard story, many, many details I can't mention them all here, because that would take a lot of pages. I just wrote the main lines about my trip.

I wish that other people somehow might find something inspiring in my adventure. And I wish that I can find peace and love and a bright future in my new homeland.

Happy birthday SVOI!

By Ingunn Westerheim

From June 3rd to 4th the Swiss OI-organization celebrated their 30th anniversary. And I was so fortunate to be invited. How grateful I am for that! Because it was a great week-end filled with interesting talks about health issues (the spine) and a wonderful celebration consisting of speeches, delicious food and wine, magic, disco and entertainment. I even got to see my boyfriend play his farewell concert with his band. I could hardly ask for more. The Swiss truly know how to celebrate and show hospitality. I felt truly welcome.

The only worry I had (and that was a big one, I can assure you), was that I had promised the Swiss president, Therese to give a speech in German. My first. I rarely get nervous from speaking in public nowadays. But this Saturday I almost suffocated from nervousness. But with help from OIFE’s secretary, a glass of chardonnay and moral support from André– it all worked out. I give my best regards to the Swiss OI-
society, and hope I will be invited when they celebrate their next big anniversary:

“I’ve been looking forward to this event for several months. First of all I found the topic of the meeting very interesting. Strengthening our backs means a lot to me as an adult with OI type III. My back and neck are probably the two factors beside my teeth, that have caused me the most problems as a person with OI. When I was younger, I used a corset for many years. And that helped with the pain that arose when I was a teenager. And here’s a tip – if you are struggling to loose weight, I can really recommend a corset. The last year I wore it, I had big problems eating anything at all. Unfortunately I also started to have problems breathing because there was so little space left. After a rib fracture I decided that my back had to do the hard job of keeping me vertical by itself.

Having a functioning spine is important. Both physically - but also in a more symbolic ways. Sometimes in life you face problems. Then it’s important to have a good spine to help you stick to the right decisions. A good spine can also give you endurance, when life gives you challenges of various kinds. Whether this is pain, fatigue, sick family members, problems at work, dating problems or just everyday challenges.

Cheers!

I have heard rumours that the Swiss OI-organisation might face some kind of crisis the next year, because too few people are willing to take over the great job that Therese has done. I encourage you all to ask yourself: What can I do for the Swiss OI-organisation? Because my belief is that the organisations STILL play an important role in peer work, in policy making, in working for research and new treatments and in creating arenas where people of all ages can meet. Young children come to OI-meetings and see that older people with OI get married, get an education and jobs. It’s all about role models. I have a strong feeling that the OI-organisations have played a key role behind the fact that so many OI-people live independent lives.”
EURORDIS & ECRD in Edinburgh

by Ingunn Westerheim

In May I had the great pleasure of visiting Scotland for the first time. The purpose of the trip was to attend the ECRD-conference and EURORDIS’ general assembly on behalf of OIFE, together with Inger-Margrethe Stavdal Paulsen (president of NFOI) and Rebecca Tvedt Skarberg (patient representative for BOND ERN).

EURORDIS is an alliance of patient organisations representing more than 700 rare disease patient organisations in more than 60 countries. Every second year EURORDIS host a big conference (ECRD) on rare diseases and orphan products together with other stakeholders in the rare disease field. This year the slogan was “Game Changers”, and one of the main topics was the establishment of European Reference Networks (ERNs), that can hopefully turn out to be a game changer for rare diseases like OI. In June 2016 an application to form an ERN for rare bone disorders (called BOND) was sent to the EU Commission. The Commission will decide about approval during 2016 and most likely BOND will be up and running from 2017. The talks about ERNs at the ECRD-conference were both informative and thought provoking. It will be interesting to see how the new network of healthcare providers (clinics and centres of expertise) will develop in the future years.

Though provoking (or maybe just provoking) was also professor Tom Shakespeare’s talk. Shakespeare himself has achondroplasia, and told us that we are disabled by society as well as our bodies, implying that the social model of defining disability is inadequate. I think he is absolutely right. Disabilities like OI are multi-factorial. We have health problems, but we also have challenges created by barriers in the society. That is why we as patient organisations have to work for better research, treatment, drugs, rehabilitation, and follow-up for people with OI. But we also have to work for access to education, mobility aids, necessary assistance and universal design. And not to forget – fight against discrimination of people with OI in all aspects. If these aspects are not taken into consideration – we end up with poor quality of life. Even if health
Before the ECRD conference, Inger-Margrethe and I took the train to Dundee to visit our friends in the Brittle Bone Society (BBS). The BBS headquarters has been situated in Dundee for many years and we were given a warm welcome by Patricia and Coreen as well as other members of the staff.

We also met some of the central volunteers (or trustees as the BBS call them). We got a tour around the BBS headquarters and got to hear about different BBS projects.

For instance their successful patient day events they have had around the UK in 2016, where they welcomed around 250 participants of all ages in Belfast, Dublin, Glasgow and Birmingham – with other events planned for Bristol and London. We also got to hear about their Wishbone day video, reaching 50.000 people and their new Research Strategy – a topic that will be discussed further at the Annual General Meeting of OIFE in Lisbon.

The BBS has also started an ambitious project to call for a fully funded Adult OI Service in the UK. So we had a lot to talk about. I was also happy to receive a gift from trustee Yvonne Grant to bring back to the OIFE as a token of the good collaboration between the BBS and the OIFE, that we hope will grow even stronger in the years to come.
Interview with OIFE’s patient representative
Rebecca TvedtSkarberg (Norway)

OIFE: In June 2016 European clinics sent an application to the EU Commission to be approved as a European Reference Network (ERN) for rare bone disorders (BOND). The patient representatives were chosen after an election process, where the OIFE suggested you as their candidate. Congratulations on becoming a "BOND-girl"! We hope you can enlighten us with some information on the ERN process.

What is a European Patient Advocacy Group (ePAG)?

Rebecca: This spring 24 applications were made to the European Commission to become European Reference Networks (ERN). An ERN is a group of health care providers (HCP) in a specific diagnostic area. The health care providers applied from all over Europe to become a part of their designated network. The European Commission felt it was essential that the patients themselves played an active role both in the establishment and the running of the networks. Therefore they set the criteria that patient organisations had to be part of the whole procedure from start to finish. EURORDIS helped making this possible by launching the idea of EPAGs - European Patient groups. EPAGs are parallel to ERNs and are to be constructed around specific diagnostic areas. Patient representatives were elected from each Epags. From the rare bone ERN (BOND) we are 3 elected patient representatives: A woman from the Portuguese achondroplasia organization, a man from the French OI organization and myself. The EPAGs role is to provide input and experience on living with a rare disorder to the ERN through the elected patient representantives. The patient representatives will be present in the organizing structure of the ERNs.
Some patient groups have already joined their assigned Epag but many are still not aware of this possibility to give input and help strengthen the ERN knowledge base. Together with EURORDIS the patient representantatives are working on a letter to all the HCP in each ERN with a request for contact details to the patient groups they are in contact with. By doing this we hope to reach even more people with rare disorders and strengthen the knowledge base of the ERNs.

**OIFE: What has been/will be your role in BOND?**

**Rebecca:** After being elected I participated in the writing process of the application. The patient representatives were added to the mailing list of the HCP applying establish BOND. There were a lot of emails back and forth before the lengthy application - finally it was ready. We are currently waiting for the EU to process the application and hope to receive an answer around Christmas time. We have received positive signals that say that all the participating HCP have met the approval by the EU. Although the ERN has not been officially established yet, EURORDIS felt we should not wait to get the contacts of the organizations and help group them with the appropriate EPAG. Therefore we have been working on a letter to HCP asking them to put us in contact with patient groups they know of.

**OIFE: There are 20 different diagnostic groups in the ERN-process and OI is placed in the group for «rare bone disorders» (BOND) - what do you think of this?**

**Rebecca:** There are 24 ERN applications around specific diagnostic areas. There is no clear cut between many of the groups because of the complexity of rare disorders. Many disorders have so many different symptoms it is almost impossible to group them. In OI we have many other complications beyond fracturing, especially as we grow older. Still the HCPs who know about OI tend to be orthopedists. These top doctors in Europe have joined together in BOND being fully aware that the rare disorders in BOND have a wide range of other complications that all play their part when living with the rare bone disorder. Also the different ERNs will be working together and exchanging knowledge to grow as a network.

**OIFE: How has the collaboration with the professionals been so far?**

**Rebecca:** Very good. They are very focused on our input and contributions. I look forward to actually working together in the network when the application is processed.

**OIFE: In your opinion – what are the most important goals/tasks for BOND?**
Rebecca: Creating awareness and getting the messages out there that some treatments are available if you have a rare bone disorder and that although rare some of our health issues are common and need treating on the same level as other patients. I also feel we have an important task of showing that in spite of rare bone disorders many of us are rated to have a high quality of life, we contribute to society and lead successful lives if we are provided with the right treatment and care.

OIFE: What will the greatest challenges be?

Rebecca: Understanding the different systems in the European countries will be a necessary challenge in order to collaborate across borders.

Interview with Dr. Sangiorgi (BONDERN)

Dr. Sangiorgi and his team sent an application in June this year to be approved as a European Reference Network (ERN) for rare bone disorders (BOND). It is up to the EU Commission to decide if the application will be approved or not.

OIFE: What is your relationship to rare bone diseases – in particular Osteogenesis Imperfecta (OI)?

Dr. Sangiorgi: Our Department has worked with hereditary rare disorders (RD) from 2003 through a multidisciplinary day-clinic and has used Disease Registries to identify the best strategy to manage fragmented information. Thanks to our IT-system, the Osteogenesis Imperfecta Registry (ROI) has allowed us to collect clinical, genetic, genealogical and imaging data on our HL7 compliant platform for more than 600 patients with OI, creating a significant dataset of information.

OIFE: BOND is divided in two - can you tell us the reason for this?

Dr. Sangiorgi: The BONDERN brings together all rare diseases (congenital, chronic and genetic) that affect cartilage, bone and dentin. This large field is divided into two main categories - skeletal dysplasia and metabolic bone diseases. These 2 categories are subdivided in several thematic and sub-thematic groups. The nosology and classification of genetic skeletal dysplasia delineated in 2015 more than 430 various
rare disorders. More than 50 specific metabolic bone diseases are described to date, but without official classification. Some disorders, such as Osteogenesis Imperfecta (OI) or Morquio disease belong to these two categories. The principles of diagnosis, management and follow-up are quite overlapping, giving a greater coherence and consistency for categorisation.

The impossibility to consider all these diseases led our group to choose 11 main thematic groups, and, among these groups, to emphasize 3 major diseases that will be prioritized, serving as a starter/template for the 2 first years: Achondroplasia, OI and X-linked hypophosphatemia. The rationale for choosing these leading diseases, and the main thematic/sub-thematic groups of disease is based on 5 central arguments:

1. disease frequency
2. severity, requiring an urgent improvement in early diagnosis and management
3. difficulty and complexity of the diagnosis, requiring a dissemination of the diagnostic expertise and modern tools;
4. difficulty and complexity in the treatment and management art, requiring also to ensure a better diffusion of symptomatic treatment or surgical techniques
5. current development of new drugs from basic research through translational research, or through biopharmaceutics research and development collaborations.

**OIFE: How many clinics are involved in BOND?**

**Dr. Sangiorgi:** The Network is composed of 38 members across 10 Member States (Belgium, Czech Republic, Estonia, France, Germany, Italy, Netherlands, Portugal, Sweden and UK).

**OIFE: What role does the patient representatives/ePAGs play?**

**Dr. Sangiorgi:** The development of the BOND network has been driven and informed by patient group (ePAG) input.

BOND will work with ePAGs in all activities to ensure patient-focused developments, with patient-reported outcome and experience measures to be adopted as specific outcomes against which to assess BOND performance in improving healthcare.

E-PAG representatives will chair and/or participate to the meetings of the following BOND Governance Boards: Steering Committee, Management Committee and Scientific Advisory Board.

**OIFE: In your opinion – what are the most important goals/tasks for BOND?**

**Dr. Sangiorgi:** BOND will bring rapid exchange of information, skills and practice to shorten time to diagnosis, and treatment. BOND will develop, with ePAGs, evidence/consensus-based guidelines to improve agreed outcomes in the 3 pilot
BOND will develop guidelines that allow outcome measurement in relation to service inputs, leading to development and dissemination of best practice. BOND will identify issues relating to care and access. Initial data suggest even “expert centres” are not fully equipped to provide integrated multidisciplinary care— and establish clear standards of care. BOND will develop patient-reported outcome and experience measures to guide these. As new therapeutics develop (anabolic therapies in OI) BOND will ensure rapid access to studies for affected patients.

**Calendar**

**2016**
September 22-25: OI-meeting adults NFOI with medical TRS-seminar
October 5th – 6th: III Congreso Internacional de Osteogenesis Imperfecta y XIII Curso Monográfico de Ortopedica Pediátrica, Hospital Infantil de México Federico Gómez
October 6-8: 1st Hispanoluso OI Congress in Lisbon, Portugal (OI in 2016)
October 6th: Meeting Skeletal Rare Diseases Coalition of Supporting Partners (IOF), Lisbon, Portugal
October 9th – 10th: OIFE AGM, Lisbon, Portugal
October 23: Medical meeting in Leuven, Belgium (ZOI – Belgium)
November 10th-11th: IOF Training Course – “Genetic Disorders of Bone and their adults expression”

**2017**
April 28th – 30th: AGM & family meeting NFOI, Oslo, Norway
May 6th: Wishbone Day
May 20th – 21st: SVOI Annual Meeting, Nottwil, Switzerland
May 26th – 28th: OIFE AGM, Poland
September 1st – 3rd: OI Austria 10 year anniversary, Wien, Austria

**2018**

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