



Newsletter Number 16 Sept 2012

Editorial

Dear friends,

preparations for "Fragile Bones – Unbreakable Spirits?", the Conference in Lisbon from Oct. 26th -28th 2012 are in full swing. We can tell you more about the program now as some speakers have confirmed. For instance:

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Editorial

**German research
award for OI**

**HPP awareness – A
disease that can
look like OI**

Report: OI Bulgaria

Did you know that...

Margarida Custodio dos Santos (Portugal): "Living with OI and the impact on families"

Carmen Almazán (Spain): "Non-affected siblings of OI children"

Catherine Potterton (UK): "The declaration of independance"

Ingunn Westerheim (Norway): "To work or not to work, that is the question"

Maria Barbero and Blanca Ostarek (Spain): "The language of disability"

Claire Hill (UK): "Quality of life in children with OI"

Anne-Miek Vroom (Netherlands): "Quality of life in adults with OI"

Luisa Barros & Margarida Custodio d.S. will offer a workshop dealing with pain

– just to name some of our speakers. For more information around the Conference please contact office@oife.org.

In spring 2012 Ute Wallentin, OIFE's president had been invited by the ESPE (a European organization of pediatric endocrinologists) to a meeting in Bulgaria. You can read her report about a wonderful experience.

"OI or not OI?" – Taco raises and gives answers to this question in his report about a disease which symptoms can be very similar to those of OI.

Hopefully you will enjoy reading this newsletter!

Stefanie Wagner

German research award for OI

The research team at the University Cologne was awarded with the "Eva-Luise and Horst Köhler research award for rare diseases 2012". The group consistent of pediatricians, (Oliver Semler, Eckhard Schönau) geneticists (Christian Netzer) and physiotherapists (Tanja Petersen) is active in research and therapy of OI people for many years.

In March 2011 they discovered the gene for the recessive OI-VI which leads to a new understanding of the underlying mechanisms in this special type. These patients don't have a collagen problem,

Student Exchange program:

Would you like to **host** an OI-student? Please write an e-mail to office@oife.org if you can help. Thank you!

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but the bone resorbing cells are too active. Therefore the patients don't have enough stable bone and they develop symptoms like classical OI-people. Due to this new understanding the researchers used a new drug in patients with OI-VI, which is approved for adults with osteoporosis. This antibody was far more effective in the children with OI-VI, than bisphosphonates were during the last years. Due to this improvements and because the new antibody can be given subcutaneously as a short injection the new project, coordinated by Oliver Semler, wants to investigate the effect of the antibody not only in patients with OI-VI but also in other types of OI. The award was given to the team by the former President of Germany Horst Köhler and his wife Eva-Luise Köhler. The award is one of the most prestigious awards in this field in Germany.



The 4 awarded scientists together with the wife of the former Federal President of Germany Eva-Luise Köhler (from left to right: Tanja Petersen, Eva-Luise Köhler, Eckhard Schönau, Oliver Semler, Christian Netzer)

"Hypophosphatasia awareness – A disease that can look like OI"

by Taco van Welzenis

"OI or no OI?" that can be the question. It sounds like a Shakespearean drama, but for some of us it is reality. For instance when a genetic test does not bring a mutation to light or when the clinical picture is a bit "odd" for OI. Some diseases can look surprisingly similar to OI.

We now ask your attention for one such example; hypophosphatasia (HPP) or "soft bones". HPP can cause bones to bent, fracture and hurt. Further symptoms may include short

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OIFE's objectives:

- *Representing its members on a European level
- *Presenting the problems and needs of people with OI to national and international organizations
- *Collecting and publishing information about OI
- *Promoting research on all aspects of OI
- *Supporting member-societies by the exchange of information and experiences

stature, teeth problems, joint pain and low muscle tone. It is very variable, different forms exist that range from a very severe disease in children to milder forms that do not start until adulthood. Mobility can be impaired, some people need a walking aid or a wheelchair, inheritance can be either dominant or recessive. HPP is a very rare disease, it is estimated to be roughly 5 times less common than OI, although the actual number of people that have HPP is unknown. Associations for HPP so far exist in only a handful of countries. Probably physicians know HPP less well than OI too.

The OIFE is now in touch with some people from the HPP community. We spoke with Gerald Brandt who is president of the German association for HPP, and he told us of a number of cases where there had been confusion between HPP and OI. One of his HPP-friends for instance had been living for over 50 years with a diagnosis of OI until last year a genetic test revealed that she in fact has a severe form of HPP. It is possible that some people with HPP still think they have OI or that they are members in OI organisations because OI is the closest "match" to their symptoms and no local HPP group exists yet.

Is it important to know if you have HPP or not? Gerald: "Yes this is very important, mainly because over the past years bisphosphonates have become something like a standard therapy for OI. Unfortunately bisphosphonates are absolutely contraindicated for HPP patients and can even lead to worsening effects like bone fractures and joint inflammations, deposition of bone mineral crystals in the kidneys – and thus even to kidney failure. On the other hand a pharmacological company is developing a bone-targeted enzyme replacement therapy to treat HPP specifically."

Then the question arises how can we differentiate between the two and how we can test for HPP? OI and HPP might look similar sometimes but they have a very different background. OI is a problem of the collagen, while the reason for HPP is a defective in the enzyme called tissue non-specific alkaline phosphatase (TNSALP), resulting in poor bone mineralization. Gerald: "The easiest way to distinguish between OI and HPP is to measure the alkaline phosphatase activity level in serum. In most people with HPP this is much lower than normal. It is also possible to do a genetic test for HPP." But there are other clues as well. For instance in HPP people will often prematurely lose entire teeth, while in OI it is more Dentinogenesis Imperfecta and fractured teeth that we see. Usually people with OI will have more fractures, but this is not an absolute indicator, some people with OI have very few fractures and some with HPP do have many. Contrary to OI, in HPP the fractures often heal with great difficulty. Children with HPP often have low muscle tone and some people with HPP may experience kidney problems as well.

Gerald: "Especially when physicians only rely on the clinical symptoms (phenotype) and forget to take the ALP level into consideration, OI and HPP can be confused. That is why we are

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Thank you!

really thankful for the opportunity to raise awareness for HPP among the worldwide OI community. Explicitly I wish to thank Ute Wallentin and Taco van Welzenis for their cooperation.”

More information on HPP can be found here:

www.hypophosphatasia.com : General information - English

www.softbones.org : US Hypophosphatasia Foundation - English

www.hpp-ev.de : Hypophosphatasie Deutschland e.V. - German

www.hypophosphatasie.com : Hypophosphatasia Europe – French



OI Bulgaria – people with OI and their doctors bring progress to their country

By Ute Wallentin

All around the world we meanwhile have about 50 national OI-associations. In 2012, as far as we know – two new ones were founded in St. Petersburg/Russia and in Bulgaria.

As OIFE-representative and speaker I had been invited by the ESPE (a European organisation of pediatric endocrinologists) to a meeting in Varna at the Black Sea in Bulgaria's south. That weekend turned into a very special and unforgettable experience I'll never forget – although it sometimes caused mixed feelings

OIFE had been in contact with people from and in Bulgaria, who were interested in OI, for about eight years already. Then came the first occasion to personally meet a young OI-doctor, Veselin Bojadjev, in October 2011 in Dubrovnik.

The first ideas about the daily life and hardships of OI-families in Bulgaria we got thanks to the German company J.S.Evro (which is the distributor of Fassier-Duval-telescopic rods for several European countries). Many years ago they had started to bring FD-rods and bisphosphonates to Bulgarian OI children and their very dedicated orthopedic surgeon and did

lots of good for some families at least – until the contact and the help stopped all of a sudden due to the unexpected sudden and tragic death of this wellknown surgeon in Bulgaria.

The second person who made us aware of the particularly difficult situation in Bulgaria years ago (after having adopted her second OI-son from a Bulgarian institution for disabled children) was Mary Peterson Suri from the USA, a known and very active medical doctor and mother of five children. She found several OI children in different Bulgarian institutions and tried for years, but with little success, to get them into standard and adequate OI treatment.

In Bulgaria the standard treatment for OI-children was and still is in many cases growth-hormone treatment instead of bisphosphonates and telescopic rodding, as in most other countries. Despite the fact that Bulgaria belongs to the European Union for some years and that cross-border-healthcare for rare disorders should already work inside the EU, in and for Bulgaria this is still not functioning and without health insurance and up-to-date OI-care the situation was pretty desperate for most OI-patients until 2011.

But then, in spring 2011, a new and better future for OI people and their families in Bulgaria took its beginning, when thanks to an ESPE fellowship program the first young endocrinologist from the Varna Childrens' Hospital passed some months in Sheffield/UK in a wellknown OI clinic with Prof. Nick Bishop and learned the modern ways of OI treatment. Right afterwards he contacted the OIFE and half a year later he was among the participants of the big International OI Conference in Croatia and the first personal contacts were established.

At the same time J.S.Evro-company had increased its attempts of spreading knowledge about telescopic rodding and the young boss of this company is still busy building up a growing network of surgeons with the most modern training and thus made telescopic rodding in OI children widely possible and better known.

So I had been invited to come to beautiful Varna in April and I was able to give a presentation to the 40 endocrinologists present.

I had been welcomed very friendly and was able to witness an important day in the local hospital for OI- and Cystic Fibrosis patients, took part in a little press conference (where the doctors, a Bulgarian OI-mom and I were interviewed for a TV-program) and met 10 OI families. They had been invited by the team of doctors lead by Violeta Iotova and Veselin Bojadijev and by a volunteer, Vania Dobrea (affected by and working for another Rare disorder) for a first family reunion and I was able to witness not only their very lively meeting, but as well the Foundation of the new Bulgarian OI association. Eli Angelova, an adult OI woman, became the first Bulgarian OI-president and soon the association was presented to the public on a facebook site.

At the family-meeting I was as well asked to give a presentation about OIFE and this was followed by many questions concerning daily life with OI in a country like Germany.

The following day I took part in the international ESPE Fellowship-meeting of pediatric endocrinologists. More than 40 participants – mainly really young and enthusiastic medical doctors and researchers – from Bulgaria, Turkey, Romania and neighbouring Balcan-countries plus speakers from Poland, Belgium, England and Germany were present.

I was able to make innumerable new contacts to very interested and dedicated doctors – most of them are as well treating considerable numbers of OI-patients.



Apart from all the new OI-contacts I really enjoyed my first stay near the Black Sea and in beautiful Bulgaria. I met so many friendly, open and really interested people everywhere, not to mention the excellent, fresh and delicious food and the lovely spring-weather after a long, cold winter.

Varna is an old and important harbour city with lots of tourism in summer and several famous white sandy beaches, a cultural center and – similar to nearby regions in Turkey or Romania a beautiful landscape and a lovely climate. But unfortunately Bulgaria still seems to be living the shadowy existence of a slightly neglected and often forgotten “stepchild of Europe”, as I got the impression.

Due to the very friendly and open hosts and many conversations I had with the people around me, I got as well the possibility to see a little bit of the backside of the European politics and economy.

And these stories I heard and some remarks made me very aware about how difficult the European situation still is in many respects, which often did not yet make it into the media and public awareness: In Bulgaria the national economy is still very weak, I could hardly find regional or national Bulgarian products in the shops – most customers are so far mainly interested in buying the Western products they missed for so many years.

No demand for national products – that means no production, no work – so many citizens are still unemployed, but without any social welfare system to stabilize and support them. Many adventurous and brave young people are constantly leaving their home country to look for work elsewhere – those who stay, are often too old, not well enough educated or sick and they can probably only survive due to their moderate expectations and needs and their small gardens.

Living in such a country with a disability is extremely difficult, of course. While I met and saw many beggars in the streets, no wheelchairs seemed to be in use – accessibility in the streets and buildings is not really an issue yet in Bulgaria. In many places I saw and even for pedestrians walking around, sudden deep holes in the walkways are a challenge, wheelchairs meet many obstacles and the hospital was friendly and full of dedicated and well-meaning professionals and volunteers, but it would certainly need some financial extra support in order to reach “European standard”.

For OI people in Bulgaria standard or even basic adequate modern OI treatment and care are still only a vague hope and far from being reality and now, as I have new friends in Bulgaria, this is hard to accept, of course and the OIFE will do as much as possible to help and change the situation.

Anyway: Bulgaria is certainly worth a visit and a longer stay and I will be back soon!

I admired the so very dedicated doctors and nurses and the volunteers from other Rare-disorders-groups who regard it as obvious that they spend lots of effort and extra time and private resources to support the new, small OI organization. The „atmosphere of departure“ and the conviction that the future shall and will be better for Bulgarian OI families gave me a lot of new motivation and hope and I am convinced that in 10 years everything will be much improved in that beautiful country!

Let's all support Bulgaria OI as much as we can – please contact us for further information

Did you know that...

...OIFE has got two new members since 2012? With Romania and Russia 25 countries form the OI Federation Europe!