



## Newsletter Number 9 Aug. 2010

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Did you know that...

## Editorial

Dear friends,

so far this summer has been extremely hot for a few weeks, at least for German conditions. Many people were complaining about it, but, to be honest, I enjoy every day without having to use thick clothes and without asking myself "should I really dare to wheel outside on icy streets doing groceries?" So I'll keep on enjoying summer...

In this second edition of the year I am happy to present an interview with Dr. Semler, a very likeable person and competent M.D., who kindly started to work voluntarily as medical website editor for OIFE.

Laurette's report about how she met a girl with OI in Chile and Anna's abstract about her final paper "O.I. in Movies" are two interesting topics in this issue, too.

Now I hope you will enjoy reading the newsletter!

Stefanie Wagner

## Interview with Dr. Oliver Semler

**Many OI-people know Dr. Semler as he has been active in the German OI-society and has presided the "OI in Motion" Conference of the OIFE. Affected by OI himself, the following interview also shows a private side.**

**OIFE: Has it always been your wish to study medicine regarding your handicap?**

**Dr. Semler:** Since early childhood medicine was always something positive for me. Normally, when I had a fracture, I arrived at hospital with a lot of pain, and after treatment, either with a cast or by surgical procedures, the pain was improved after a few days. So after some years the idea of becoming a doctor myself grew more and more. After I finished school I was thinking what to do now. My parents studied law (one is a judge, one a lawyer) so this was not something totally out of sight for me. But nonetheless I decided to do something on my own and so it was an easy decision to study medicine.

**OIFE: How big was the challenge for you as a student of medicine with OI?**

**Dr. Semler:** Being affected by OI as a medical student has some special aspects. Luckily I can walk without any walking aids quite good, but sometimes it was not so easy to arrive in the right room at

the right time. While others were using their bicycle I had to carry all my books and had to walk. So studying with a handicap needs some more planning how to arrive where you are supposed to be.

Another problem was, that there are some (but luckily only a very few) topics, where you need physical strength. Like in Emergency medicine, where you learn how to carry a patient out of a dangerous situation etc. These courses were always a bit problematic, but in the end, sometimes after a few discussions with my professors, somehow I managed these courses.

The other point being affected by OI is of course, that you are always something special, everybody is shy, has no idea if he can ask why I am looking different and so on. That was something that happened to me a lot, but I don't think it is related to the medical courses. It is related to general behavior of people and I am sure, I would have had to fight the same questions and kidding in any other field as well.

## Projects:

[Making-friends  
Project](#)

[International OIFE  
youth weekend](#)

[Student Exchange  
Program](#)

[OIFE Pass](#)

### **OIFE: Can you describe the topic of your M.D. thesis?**

**Dr. Semler:** I did my M.D. thesis in the hospital where I "grew up". By that time surgical treatment was completely different to the treatment today, because there were no telescopic rods at that time. Because of this a lot of other orthopedic materials were used to stabilize the bone and to treat and prevent fractures. My thesis was to look at effects and complications of different surgical strategies used in patients with OI in this special hospital.

### **OIFE: You have been working at Shriners Hospital in Canada. Can you describe your tasks there?**

**Dr. Semler:** In 2009 I spent 4 months for a research project, sponsored by the German Society of pediatricians, at the Shriners Hospital in Montreal, where Bisphosphonates were first used in the early 1990s. My first research project was to look at the necessity of calcium and vitamin D supplementation in children with OI, treated with pamidronate. It turned out that this project was difficult to realize due to a not standardized therapeutic regime. So I added another project and looked at x-rays of the skull, which are done routinely at start of therapy in this hospital. I was looking for abnormalities of the skull bones (wormian bones) which are often described as something very typical for OI. Additionally this is a parameter which is sometimes used as one parameter, when differentiating between OI and child abuse. The result was that wormian bones occurred more frequently in severely affected children, and in less than 50% in type I persons. Additionally there is a correlation with some typical genetic mutations.

### **OIFE: You work as pediatrician. Can you describe your work?**

**Dr. Semler:** I am running an outpatient department for children with different bone diseases predominantly OI. I am responsible for the medical treatment (Bisphosphonates – in Cologne we use neridronate) and I am also responsible for the OI patients who participate in our rehabilitation program. At the moment I am trying to build up a surgical center together with our orthopedic department for children with OI using the Fassier-Duval-rod. This means, more or less, that I am responsible for the coordination of the whole medical treatment regime for our OI patients, not only for kids but for adults

## News in Brief

OIFE got a lot of positive feedback on **OI in Motion**, OIFE's conference in November 2009. One clinic even changed its physiotherapeutical concept!

[Please click here](#) to read and watch: "a **mother-and-daughter experience of brittle bone disease**" report and video interview with OIFE's president Ute Wallentin

Please visit the **OI in Motion picture gallery** on our website: [www.oife.org](http://www.oife.org)

The **Topical Meeting for surgeons** organized by the University La Sapienza in May 2010 in Rome, Italy, was a big success. Many of the participants were from Italy.

The **OI-association Venezuela** has got a new website: [www.oicomunidadosea.org.ve](http://www.oicomunidadosea.org.ve)

**Youth Weekend** Nov. 4-8 in Belgium, last Call! Please contact: [jongeren@zoi.be](mailto:jongeren@zoi.be)

also.

### **OIFE: How can your little patients benefit from the fact that you have OI by yourself?**

**Dr. Semler:** Probably the parents benefit more, than the children benefit from this fact. The most common situation in my clinic is, that the child is affected and the parents are not. This means the parents don't know much about OI, when they come to my clinic and they are anxious about the future of their child. When they meet me, it is for many parents the first time they see a person with OI can become old, have a qualified job and live an independent life.

For the children it is easier to trust a doctor with a small size and when they grew older a lot of them want to become medical doctors themselves – I don't know why ☺

### **OIFE: When and how did you know about OIFE for the first time and what do you think about OIFE?**

**Dr. Semler:** I am active in the German OI society for many years and I think that it is crucial that affected persons benefit from the experiences of other affected people. Because OI is a rare disorder it is obvious that such a cooperation cannot be limited to national borders.

### **OIFE: How do you like to spend your free time?**

**Dr. Semler:** In my free time I like to learn something about different people and cultures. That's the reason why I like to travel. I started this during my final year a medschool when I spent 4 months in South Africa in a hospital in Cape Town and 4 months in Sydney. Since then I try to travel a lot in my free time. For me it is extremely interesting to see how other cultures look at "life" and it widens your mind if you see the circumstances in other countries.

### **OIFE: Can you tell us about one of the proudest moments in your life?**

**Dr. Semler:** It is hard to pinpoint one special moment, but I am always proud, when I have the chance to present a talk at an international conference on the topic of OI. I am proud that I am able to stand there, being accepted by the scientific community and I am also proud to act there as a representative for all people with OI.

### **OIFE: What do you think is one of the most important medical challenges in the future for people affected by OI?**

**Dr. Semler:** In the next years there will be two major challenges in my opinion. One is that different treatment strategies have to be more coordinated. For example: surgical procedures and rehabilitation programs have to work hand in hand and orthosis have to be adjusted to these new situations.

The other thing is that there will be more and more knowledge about the genetic background. This means that you might be able to give a better prognosis for an individual patient when knowing the type of mutation causing the OI. This would be very helpful for counseling parents and patients and can improve therapy if you have a proper idea about the future course of the disease.

## Student Exchange program:

Would you like to host an OI-student? Please [contact OIFE](#) if you can help. Thank you!

**Ideas?  
Feedback?  
Questions?  
News?  
Interesting stories to tell?  
Please contact the [editor](#)**

**OIFE:** Thank you for this interview!

**Dr. Semler:** Thank you!

## Report: Young woman with OI from France meets OI girl in Chile

By Laurette Paravano

The following report once more shows the importance of a project OIFE will focus on in the future: Student Exchange. Like many young people, with or without a disability, Laurette, a young woman with OI from France, had the wish to study abroad. During her stay she met Camila, a girl also affected by OI. This is her story.

"A few years ago, as a student I had the opportunity to stay one whole year in South America, in Chile to be more precise. I had always been fascinated by that strange shaped country, a long and narrow piece of land between the Andes and Pacific Ocean.

I was 21 and had some very serious reasons to go there: discover another culture, speak Spanish fluently, study new materials... But of course, those weren't the true motivations. The main reason was to prove that in spite of being fragile, small and walking with crutches, I was able to manage life by myself twelve thousand kilometres away from my home country.

I think that I didn't really consider it as a challenge for myself, because I was quite self confident and didn't worry about how to manage there. Maybe I can say thank you to my parents who never imposed unnecessary limits on me and encouraged me to do much, to dare living every experience as far as possible. But I just needed a proof for the others, to be recognized as someone who was able to manage, to be independent... I can say that this experience helped me a lot when I had my first interviews to find a job!

My stay in Chile has been very pleasant and interesting. Taking the bus was quite a dangerous sport but I could do it. People are very kind, helpful and uncomplicated towards disabled persons.

But there's another story within my story: In Santiago I met a young OI girl, Camila. She was 12 years old, had a very severe form of OI and had fractures almost every week. Camila was from a poor family, but she was quite lucky compared to other OI or disabled children: she had her own wheelchair and was studying in a college. Her Dad took her to college or to hospital by car. Now she is grown up and has had no more fractures. She is studying graphic design and is very happy with it.

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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
- [www.oife.org](http://www.oife.org)



Camila and Laurette (behind)

However, only very few disabled children have a chance like Camila, and I could see the difficulties poor families with disabled children had to face. They live in poor houses and cannot heat properly in winter, so the children often have a lot of respiratory problems. Additionally, there is smog in Santiago. Some children don't have a wheelchair and cannot be treated in hospital because their parents don't have a car and can't afford a taxi. They cannot get regular therapy, and sometimes they cannot afford to buy medicine.

They also have problems filling in forms or understanding the mail they receive from authorities etc. Some single mothers have to leave their child at home while they go to work. Many children even do not get a basic care and just "survive" somehow.

Camila's Mum, called Popa, took great care of families with disabled children in her neighbourhood. Together they formed a group, working hard to get support so they could build the "Welcome house". Here they take care of 40 children with various and partly severe disabilities every day. The children receive food, basic therapy and they do some workshops. The "Welcome house" is financed by donations. For readers who also speak Spanish, please find more info here: <http://amigosdejesus.cl/>

## "OI in Movies"

Abstract by Anna Rossi

**In the last newsletter edition Anna, a young OI woman, introduced herself as the new OIFE Youth-coordinator. Meanwhile she finished her studies and got the bachelor's degree in Communication Sciences. Her final paper deals with Osteogenesis Imperfecta in Movies. The abstract as follows:**

For centuries, a lid has been clamped on the social situation of disabled people, who have been confused with, lumped together or recognised as simply one of many other marginalized groups of outsiders. Only recently, have they begun to gain importance in the social and governmental debate. The story of their integration has been both slow and long and is still not yet complete. The relationship with diversity is, to date, one of the most difficult problems that humanity has had to face: over the ages, in different cultures, diversity has been seen as threat, as something to be frightened of, a direct attack on our identity. The cinema, as a mirror

of socio-cultural development and change, could not remain indifferent to the social problems that disability harbours. In its representations of these, the cinema has alternated different points of view; thus, alongside pitiful, compassionate, sad and poignant stories we also find frank and disillusioned ones that show up the many problems, obstacles and injustices disabled people have to face in their daily lives.

The present paper offers an analysis of the ways in which the cinema has been able to represent Osteogenesis Imperfecta, an hereditary pathology of the connective tissue which affects one person per 10-20,000 and is characterized by fragile bones that break easily. Because of this extreme fragility it is also known as "brittle bone disease" or as "the glass-bones/crystal-bones disease". Someone born with this disorder is affected by it throughout his or her whole life, and suffers many fractures, often due to very slight traumas and ordinary reactions such as a scare, a laugh or a cough. In addition to fractures, people with OI often suffer from many other problems -and not only physical ones- linked to the pathology; these are portrayed more or less frequently and correctly in the four movies taken here into consideration for the analysis: *Unbreakable* by M. Night Shayamalan, *Le Fableux Destin de Amelie Poulain* by Jean Pierre Jeunet, *Fragile* by Jaime Balaguerò and *The Sixth Happiness* by Waris Hussein.

In *Unbreakable* O.I. is seen as the emblem of fragility, clearly defined through the sharp contrast with its opposite: the invincibility of the hero David Dunn throws into relief the fragile Elijah. Throughout the movie the latter is convinced that the only way he can give sense to his poor-health and fragile existence is by proving that someone else is invincible and indestructible. Only this can give his life meaning. For Elijah O.I. becomes a sort of way towards the discovery of his place in the world. Whereas, in *The Fableux Destin de Amelie Poulain*, the character played by Serge Merlin is an artist, a wise man: this places him in conditions that presuppose a certain degree of concentration and isolation which, in effect, becomes absolute for the old painter Raimond Dufayel. He lives in his totally isolated and muffled flat, safe in his cocooned "nest". Despite this, he is immensely fascinated by the world and its life. Raimond's physical fragility keeps pace with Amelie's psychological frailty; through their conversations, incidentally about the girl who hides behind a glass, they become friends and begin to give one another advices. The most important of these is Raimond's last advice that convinces the young girl to start living and leave behind her fear of colliding with life's emotions and experiences. By living her life fully she will somehow also give the old painter the opportunity of enjoying the feelings and the joys of life.

Instead, in *Fragile* O.I. is a dark, mysterious, and terrifying presence; it brings unexpected and astounding pain and suffering to Mercy Fall's little patients. Fear of pain finds in the nightmarish tension of this horror movie its finest transposition. This aspect is, unfortunately, a sad reality for those affected by this pathology.

*Sixth Happiness'* representation, on the other hand, is more realistic. This film tells the story of Brit but is actually based on Firdaus Kanga's autobiographical novel *Trying to Grow*. From the most banal, unpredictable and inexplicable fractures of his childhood,

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**Thank you!**

the author and his fictional alter-ego Brit, learn how to handle their fragile lives and how to live with their repeated, multiple fractures due not only to the pathology but often simply to life and its various troubles.

In each of the movies considered many real, typical elements and aspects of the pathology and of the daily lives of those affected by it are brought to light. It is also true that the technical requirements of the cinematic transposition has, of itself, caused some of these aspects to be, at times, altered or exaggerated; this is why, in the end, they may appear less reliable.

## Rare Disease Day 2010 - Results

**In the last edition there was a hint to the Rare Disease Day 2010 which took place on February 28<sup>th</sup>. You can read some of the results in the following article.**

The third edition of Rare Disease Day, organised by EURORDIS and its Council of National Alliances, was a resounding success. Throughout the world, a multitude of events took place on and around February 28th to raise awareness of rare diseases and the millions of people affected by them. « Rare Disease Day grows in strength and quality every year, » says Yann Le Cam, EURORDIS CEO. « This year the campaign has made one step further towards international stature with new partners from as far afield as Japan and Brazil. The campaign has been impressive not only in terms of numbers but also in terms of quality and creativeness ». More information see: <http://www.eurordis.org/content/rare-disease-day-2010>

## Did you know that...

...you can still participate in the **OIFE photo contest**? A **first prize of 100 Euros or an MP3 player** and a second prize of 50 Euros will be awarded for the best pictures. Full rules and contest forms can be found on the OIFE website under „Projects“. So grab your camera and take a shot! Good luck!