OSTEOGENESIS IMPERFECTA
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*Represents their members on a European level
*Presents the problems and needs of people with OI to national and international organizations
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New OIFE Youth-coordinator

We welcome the new youth-coordinator Anne-Miek from The Netherlands! Here is her introduction:
"Recently I was informed by Taco van Welzenis (the former youth-coordinator and now vice-president of the OIFE) about a vacancy for Youth Coordinator within the OIFE. Because of my (pleasant) experiences in the VOI (the Dutch OI society) I told that I would be happy to do this job. I am already Youth Coordinator in the VOI. We have a very enthusiastic team of young people. I also represent the youth as a general member on the VOI board. In the Netherlands we so far had three OI-youth-weekends (one of them international). Besides that we organised a lot of nice single day-activities. I would like to use my experience to help expand the contact between young OI people in Europe.

To introduce myself; my name is Anne-Miek Vroom, I'm 25 years old. I live and study in Groningen, a city in the North of the Netherlands. I just finished my bachelor in Sociology and started the master Clinical and Health Psychology at Groningen University. I had about 60 fractures (OI type IV), my back gives me most problems. That is why I live on my own in the middle of the week and relax at my parents' home during the weekend. In my spare time I love to spend time with my family, friends and two dogs. In the time ahead I hope to make myself familiar with all Youth & OIFE matters. If you want to contact me you can use this e-mail address: youth-coordinator@oife.org

I am looking forward to meet you! Anne-Miek"

News from the Making Friends Project

Making Friends is a free OIFE project with the goal of bringing individual OI people and their families together. The program is used for instance to find chat buddies, pen friends or people to visit abroad. Some people want to exchange experiences about treatments, want to learn what life with OI in a foreign country is like or seek tips for travelling.

At the moment there are 54 participants from 23 different countries. Most participants have OI themselves, but also parents, partners and other family members take part. It's a project for all ages, the youngest participant is 7 years young and the oldest is 65.

A big update is scheduled for January 2007, when a new contact list with the names of all participants will be made. We strive for quality rather than quantity. This means that every now and then we will ask participants to confirm their wish to stay in the project. Those people who do not respond are automatically excluded form the next contact list. This might seem a bit harsh but it is done to ensure that the contact list doesn't clog up with people who have lost interest or who are still in our system with an old e-mail address for instance. For more information and online registration as well as registration forms in languages other than English visit: www.oife.org/makingfriends.html

OIFE became member of EURORDIS in 2006

Since October 2006 OIFE is a member of EURORDIS (European Organization for Rare Diseases). It is a patient-driven alliance of patient organisations and individuals active in the field of rare diseases. Eurordis' mission is to build a strong pan-European community of patient organisations and people living with rare diseases, to be their voice at the European level, and - directly or indirectly - to fight against the impact of rare diseases on their lives. (See the website www.eurordis.org for further information.)

OIFE President Ute Wallentin has spoken to EURORDIS officials about the problem in Romania that OI-children would not get treatment for free anymore. She was asking for their support and EURORDIS reacted immediately by writing two letters to the Ministry in Romania and to the main hospital there.

Meanwhile the OI-children in at least parts of Romania get the treatment again for free - and our contacts there have got much support by a national umbrella organization for rare diseases.

New OI-gene discovered

In recent years a lot has been discovered about the genetic background of OI. Numerous changes in the DNA have been described. These are called mutations. So far all OI mutations have been found in genes that code for collagen; COL1A1 and COL1A2.

However it also became evident that not all people with OI have mutations in these genes. Scientists estimate that mutations in COL1A1 and COL1A2 account for 85-90% of all OI cases. This figure leaves a considerable subset of OI people that must have a different cause

for their OI.

The COL1A1 and COL1A2 genes code for collagen, or to be more precise for two collagen building blocks called procollagens. In the body procollagen is modified and three procollagen molecules are assembled into one collagen molecule. Scientists speculated that when something goes wrong in the modifying process that this could also lead to OI. In other words the collagen genes function normal but the resulting procollagen is not processed in the right manner so you still end up with a collagen disease. So far however there was no evidence to back this theory up.

Many different substances act in the modifying process. A crucial substance has now been identified, as well as the gene responsible for it. This gene is the CRTAP-gene. Researchers were able to demonstrate that mutations in the CRTAP-gene affect the modification process. Both in humans and mice this was leading to forms of OI1. Interestingly so far all CRTAP related cases of OI are recessively inherited, this in contrast to most cases of OI which are inherited in a dominant fashion.

What is the importance of this find for people with OI? The fact that a completely different pathway has been discovered that leads to recessive forms of OI can have implications for such things as genetic counselling, genetic testing and perhaps even treatment. For instance it is not known if people with CRTAP related forms of OI respond in the same way to treatment as people with collagen gene related forms. Also there could be some differences in symptoms that so far have gone unnoticed. Perhaps even more OI genes will be found in the future that also play a role in the modifying process. It is encouraging to see that major scientific breakthroughs still happen in the field of OI genetics.

1Lee, B. et al. Cell Vol. 127, 291-304, October 20, 2006

http://www.cell.com/content/article/abstract?uid=PIIS0092867406012153&highlight=crtap

New OI clinic in Moscow

Dr. Natalia Belova reports:
“We finally have organized an OI clinic in Moscow. It is based in the American Medical Center, where I have position of Head of Pediatric Dept. I have OI clinic every week. Now I have about 7-10 NEW patients from all former USSR every month, because our Center is by far the only place in Russia, where patients with OI can get treatment with Pamidronate. (You may find some additional information about the Clinic on our website www.amcenter.ru - see English version).

I guess that we probably have one of the biggest OI society - for 20 years of experience there are more than 600 patients with OI - both children and adults. I still communicate with my former patients and their families. We also organized in our Center a Summer Birthday Party, and invited all OI patients from Moscow area . It was fun, and probably the first entertainment for our OI kids. By far not all children receive treatment in the clinic free of charge.

The most important thing is that we started to work together with Georgean OI society. I am just back from Tbilisi, where we were discussing plans for future. Khatuna Saganelidze is a tremendous help for our new OI society, and we have planned many things for our mutual work during the next year.”

News in brief:

Padrinos-OI: the official foundation (and legal registration) of a new godparents-association in Germany has been realized. It is a non-profit organisation with the objective to support individual OI-people and their families whenever their care is not possible by other means.
Philippines: During the last weeks several families contacted OIFE for advice regarding OI. We are glad to know a geneticist from Cebu, Philippines, who herself has contact to several medical centres in the country. She is very good taking care of the families.