Soft Tissues & Soft Issues
OI in adults is more than fractures

Topical Meeting on Osteogenesis Imperfecta
Oslo, Norway 2015
“OI is more than fractures”
Foreword from the President of OIFE

As you know, osteogenesis imperfecta (OI) is in most cases no bone disease, but a general collagen disease. I have lived with it for so many years and live well, so I don’t really want to call it “disorder” or “disease”. But when the often inappropriate name “OI” was given to this complex and sometimes fascinating “form of being”, its complexity was not yet understood.

Osteogenesis imperfecta was and is falsely reduced to “brittle bones”. And most professionals and families first care for and concentrate on the treatment of painful fractures, which especially children suffer from. Fortunately with age and more caution, fractures diminish. And since successful methods for treatment of children have been found, fracture rate in general gets less of an "issue", especially after puberty.

But then nature does not spare adults with OI. The normal aging process happens earlier and often stronger in people with OI than in the normal population. The aging process brings the neglected other symptoms of every collagen defect to broad daylight. As fractures lessen, pain increase, muscles and tendons are easily overexerted from too much stress. Also bad posture in walking patients due to scoliosis or deformities and wrong positions in wheelchairs or sitting positions (partially to avoid pain) can cause problems.

Lack of or bad quality of collagen influences as well endurance and function of blood vessels and almost all the soft tissue in the body. OI-people in general (my antithesis) show these non-fracture-related symptoms from early on in their lives. But fractures prevail and children mainly associate or define pain with "fracture-trauma". Because of this it took decades until we; adult OI-people and our doctor friends, found time to take all the side-effects of OI more seriously.

Hearing impairment, tendonitis in every part of our bodies (mainly in arms and legs), ruptures of muscles and so on, do often torment adult OI-people more that the almost forgotten childhood fractures have done. Pain isolates us, is invisible and is often not taken seriously. The high incidence and occurrence of highly painful and first invisible tissue-problems was longtime ignored and neglected by all of us. Maybe we did not regard it as “real pain”?

However – most OI-people fortunately get pretty old and have strong wishes to continue with their active and good lives. But these ignored challenges brought the decision to make soft tissue the theme for our seminar/topical meeting and to start looking deeper into these "other", but very important aspects of OI.

Thank you to all those who participate! And also thank you to Ingunn Westerheim - the OIFE’s coming president, and her team for its preparations!

I’m wishing you all good, inspiring and fruitful discussions in Oslo!

Ute Wallentin
OIFE-president (for a few more days)
“What are the real issues?”
Foreword from the local organizers

At OI conferences we usually hear a lot about genetics, orthopedics and bisphosphonates. The focus is often on treatment strategies for children with OI. But what happens when the child grows up, fractures less and the pediatrician is no longer around?

One and a half year ago I was attending the European Conference on Rare Diseases in Berlin together with Ute Wallentin, who sadly could not attend our seminar because of a fracture. We talked about a possible theme for the OIFE’s topical meeting in 2015. And suddenly an idea popped into my head. What if we called the seminar ‘Soft Tissues & Soft Issues’ and focused on adults with OI and their non-skeletal problems?

Because what is really the main challenges for adults with OI? It’s not the fractures anymore. No, it’s the everyday pain and lack of energy that everyone is complaining about. Some of it might be bone pain. But a lot of it is due to challenges related to muscles, tendons and ligaments. And why are we so exhausted all the time? Maybe our poor collagen has other effects than those we have focused on so far? And how many people with OI do really have lung-, heart or gastric issues? And what can we do to prevent these problems?

Unfortunately few professionals know a lot about these issues. And why is that? Perhaps it is caused by the fact that adults with OI have few or none interdisciplinary clinics or services, like children do? When you turn 18 you change from being a whole body and person to a lot of body parts belonging to loads of different specialities. Very few doctors have seen a lot of adults with OI in their clinic. And the GP will never ever really get it, will he?

So we asked dr. Lena Lande Wekre, what she thought about the idea – and she was immediately positive: “This is exactly what many of the adults who come to the TRS centre is talking to us about. But there is very little research and knowledge about OI and soft tissue. Maybe such a seminar could spark an interest and create more research?”

Since the OIFE is not the only OI-umbrella hosting topical meetings on OI, we decided to make it a joint effort with OI-Norden and the Norwegian OI-association NFOI. The program committee has had representatives from all three organizations, and I would hereby like to thank Kis, Taco and Rune for their effort and advice. Also I would like to give a special thank you to Inger-Margrethe, my vice-president and expert list keeper. Without you this would not be possible! And also a big thank you to all the other volunteers who have helped us on different tasks.

What we were mostly worried about when we started this project, was if we would find any lecturers. Was there any interest out there for the topics? Could we get in touch with professionals who had experience with adults and OI? Or were we going to be stuck with a lot of participants but no professional contributors? Our nightmare did fortunately not come true. Around 80 participants from 21 different countries are attending. And we have a good mix of people who have OI themselves and doctors, nurses, OTs, PTs, psychologists and several other types of professionals.

Oslo, Norway 2015
And a big thank you to all the professionals who have agreed to come here to talk, with little or no financial support. Without your sporty attitude, three volunteer organizations would not be able to pull this off. And we are really happy to see that you are interested in adults as well.

I am looking forward to two days where we will focus on issues that we have talked a lot about before – but not in a seminar setting like this. Perhaps the seminar will leave us with more questions than answers, but we hope it will spark an interest in the matter and stimulate further research.

Ingunn Westerheim
Project manager “Soft Tissues & Soft Issues” and president of NFOI

ORGANISERS

Both the Osteogenesis Imperfecta Federation Europe (OIFE) and OI-Norden usually have a topical meeting about osteogenesis imperfecta every 2-3 years. This time it has been a collaboration between the two umbrella organizations and the Norwegian Association for Osteogenesis Imperfecta (NFOI). Most of the practical preparations have been done by members of the Norwegian board.

PROGRAM COMMITTEE

NFOI
Ingunn Westerheim
Inger-Margrethe Stavdal Paulsen

OI-Norden
Kis Holm Laursen
Rune Bang Mogensen

OIFE
Taco van Welzenis

We have also asked the Medical advisory board of NFOI and TRS National Resource Centre for Rare Disorders for advice during the process.
SPONSORS AND OTHER SUPPORTERS

We send a big thank you to all people who have contributed with the program and practical preparations!

We are extremely grateful for the financial support of Alexion, Studieforbundet Funkis and the support from the different lecturers regarding travel costs and fees. Without this support, three volunteer organizations with limited resources would not be able to host such a meeting with participation from both professionals and people with OI.

The professional contributors

Lena Lande Wekre, MD, PHD (Norway)
Michael Bober, MD, PHD (US)
Olga de Vries, PT (Norway)
Lars Folkestad, MD (Denmark)
Francis Glorieux, MD, PHD (Canada)
Zoran Radunovic, MD (Norway)
Trine Bathen, OT (Norway)
Elise Christensen, social pedagogue (Norway)
Tamara Fernandez, psychologist (Spain)
Arjan Harsevoort - nurse practitioner (The Netherlands)
Ann Bett Kirkebæk - consultant physical exercise (Denmark)
Jannie Dahl Hald, MD, PHD (Denmark)
Ana Bueno, MD (Spain)
Fleur van Dijk, MD, PHD (The Netherlands)
Lydia Forestier-Zhang, MD (United Kingdom)
Harald Schubert - general manager (Germany)
Antonella Lo Mauro - biomechanical engineer
Susan Stewart, nurse specialist (United Kingdom)
Alessandra Ciliberti – psychologist (Italy)
Svein Otto Fredwall, MD (Norway)
Other contributors
Ingunn Westerheim (Norway)
Inger-Margrethe Paulsen (Norway)
Kis Holm Laursen (Denmark)
Rune Bang Mogensen (Denmark)
Maria Barbero (Spain)
Olga Wittauer (Finland/Germany)
Patricia Knötzsch (Germany)
Mads Haldrup (Denmark)
Snjezana Jolic (Serbia)
Renata Alic (Croatia)
Andreas Henden (Norway) – technical support
Vibeke Bakken (Norway) – editor of abstract book

Other supporters
TRS National Resource Centre for Rare Disorders

Financial sponsors

The organizations
Norwegian Association for Osteogenesis Imperfecta (NFOI)
Osteogenesis Imperfecta Federation Europe (OIFE)
OI-Norden

Other financial sponsors
Alexion
Studieforbundet FUNKIS
PROGRAM
Soft Tissues & Soft Issues – OI in adults is more than fractures

Wednesday Sep 16th

17.00  Registration opens

20.00  Welcome drink in the reception bar

20.30  Dinner for those who arrive Wednesday
**Thursday Sep 17th**

**09.00 Registration and coffee**

**10.00 WHAT ARE THE ISSUES?**

*Moderator: Maria Barbero*

<table>
<thead>
<tr>
<th>Time</th>
<th>Topic</th>
<th>Presenter(s)</th>
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</thead>
<tbody>
<tr>
<td>10.00</td>
<td>Welcome address from the organizing committee</td>
<td>Ingunn Westerheim &amp; Inger-Margrethe S. Paulsen</td>
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<tr>
<td>10.10</td>
<td>Short testimonies</td>
<td>Adults with OI</td>
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<tr>
<td>10.20</td>
<td>What do we know about adults with osteogenesis imperfecta in Denmark - the non-skeletal issues, eyes and quality of life</td>
<td>Jannie Dahl Hald</td>
</tr>
<tr>
<td>10.40</td>
<td>What do we know about the adults with OI in the Netherlands?</td>
<td>Arjan Harsevoort &amp; Fleur van Dijk</td>
</tr>
<tr>
<td>11.00</td>
<td><strong>Coffee break</strong></td>
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<tr>
<td>11.10</td>
<td>Joint Laxity and muscle strength in OI</td>
<td>Olga de Vries</td>
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<td></td>
<td>Possible impact on activity and participation</td>
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<tr>
<td>11.25</td>
<td>Joint Laxity in Osteogenesis Imperfecta and Skeletal Dysplasias</td>
<td>Michael Bober</td>
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<tr>
<td>11.40</td>
<td>The foot in OI</td>
<td>Ana Bueno</td>
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<tr>
<td>11.50</td>
<td>Alterations of the axes of the lower extremities</td>
<td>Ana Bueno</td>
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<td>12.00</td>
<td>Bone – Muscle unit and Neuromuscular Function in relation to Mobility</td>
<td>Harald Schubert</td>
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<tr>
<td>12.30</td>
<td><strong>PANEL DISCUSSION – SOFT TISSUE</strong></td>
<td>Moderator: Kis Holm Laursen, Olga de Vries, Harald Schubert, Michael Bober, Francis Glorieux, Ana Bueno</td>
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<tr>
<td>13.00</td>
<td><strong>Buffet lunch</strong></td>
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14.30 WHAT ARE THE ISSUES (Continues)?

Moderator: Kis Holm Laursen

Are there other issues than those of muscles and joints? Which consequences do they have?

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Speaker</th>
</tr>
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<tbody>
<tr>
<td>14.30</td>
<td>Short testimonies</td>
<td>Adults with OI</td>
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<tr>
<td>14.45</td>
<td>Rib cage deformities alter respiratory muscle action and chest wall function in patients with severe OI</td>
<td>Antonella Io Mauro</td>
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<tr>
<td>15.00</td>
<td>Life span and causes of death in OI</td>
<td>Lars Folkestad</td>
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<td>15.15</td>
<td>Coffee break</td>
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15.30 WHAT ARE THE CONSEQUENCES?

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Speaker</th>
</tr>
</thead>
<tbody>
<tr>
<td>15.30</td>
<td>1) Chronic pain and OI 2) Pain Less – OI-youngsters take control</td>
<td>Tamara Fernandez</td>
</tr>
<tr>
<td>15.45</td>
<td>Fatigue in adults with OI and other connective tissue disorders</td>
<td>Trine Bathen</td>
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<tr>
<td>16.05</td>
<td>Perfectly normal - or The Odd One Out?</td>
<td>Elise Christensen</td>
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<tr>
<td>16.20</td>
<td>Coping strategies in adults with OI</td>
<td>Alessandra Ciliberti</td>
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<tr>
<td>16.45</td>
<td>Coffee &amp; waffles</td>
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<tr>
<td>17.05</td>
<td>PANEL – ASK THE PERSON WITH OI</td>
<td>Moderator: Kis H. Laursen</td>
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<td></td>
<td>Panel of adults give examples of the challenges and answer questions from professionals.</td>
<td>Rune Bang Mogensen</td>
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<td>Mads Haldrup</td>
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<td>Snjezana Jolic</td>
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<td>Patricia Knötzsch</td>
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<td>Andreas Henden</td>
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<td>17.50</td>
<td>Exercise as pain relief for adults with OI</td>
<td>Ann Bett Kirkebæk</td>
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<td>18.10</td>
<td>Practical exercise</td>
<td>Ann Bett Kirkebæk</td>
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<tr>
<td>18.20</td>
<td>Program ends</td>
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<td>19.15</td>
<td>Pre dinner drink – in the bar (2nd floor)</td>
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<td>20.00</td>
<td>Conference dinner (3 course) – hotel restaurant</td>
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Oslo, Norway 2015
Friday Sep 18th

08.00 Registration and coffee

09.00 WHAT CAN WE DO TO UNDERSTAND MORE?
Moderator: Kis Holm Laursen

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Presenter</th>
</tr>
</thead>
<tbody>
<tr>
<td>09.00</td>
<td>Registries, research and collaboration</td>
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<td></td>
<td>The Brittle Bone Disorder Rare Disease Clinical Research Consortium (and contact registry)</td>
<td>Ingunn Westerheim</td>
</tr>
<tr>
<td>09.10</td>
<td>The RUDY-study - a novel approach to patient-driven research</td>
<td>Lydia Forestier-Zhang</td>
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<tr>
<td>09.30</td>
<td>OPEN DISCUSSION - COLLABORATION</td>
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<tr>
<td></td>
<td>How can we collaborate more regarding research and registries - between countries, between professionals &amp; between organizations and professionals.</td>
<td>Moderator: Kis H. Laursen</td>
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<tr>
<td>10.00</td>
<td>Coffee break</td>
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10.15 WHAT KIND OF SERVICES DO ADULTS HAVE TODAY?

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<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Presenter</th>
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</thead>
<tbody>
<tr>
<td>10.15</td>
<td>Adult care in Denmark – what did the adult study teach us about the need for follow-up?</td>
<td>Jannie Dahl Hald</td>
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<tr>
<td>10.25</td>
<td>West Midlands Adult OI Service - Need and Service Provision</td>
<td>Susan Stewart (via Skype)</td>
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<tr>
<td>10.40</td>
<td>Adult care in the Netherlands (Isala clinic)</td>
<td>Arjan Harsevoort</td>
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<td>10.50</td>
<td>Adult care – what do we want? What do we need?</td>
<td>Ingunn Westerheim</td>
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<tr>
<td>11.10</td>
<td>PANEL DISCUSSION – ADULT SERVICES</td>
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<td>Should adults continue in pediatric ward or should they have their own clinics?</td>
<td>Moderator: Kis H. Laursen</td>
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<td></td>
<td>What professionals should work in an adult clinic? How do we create good transition models?</td>
<td>Ingunn Westerheim, Arjan Harsevoort, Svein Otto Fredwall, Lars Folkestad, TBA</td>
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12.00 Buffet lunch
10

13.15 WHY AND HOW SHOULD WE FOLLOW-UP?

Moderator: Maria Barbero

Can better follow-up routines prevent some of the issues/complications which adults with OI face? What parts of the body should be monitored - by whom and how often?

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<thead>
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<th>Time</th>
<th>Presentation</th>
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</tr>
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<tbody>
<tr>
<td>13.15</td>
<td>Follow-up routines of adults with OI - Does age make any differences to the need of care?</td>
<td>Lena Lande Wekre</td>
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<tr>
<td>14.30</td>
<td>Follow-up of heart issues</td>
<td>Zoran Radunovic</td>
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<tr>
<td>14.50</td>
<td><strong>PANEL DISCUSSION – FOLLOW-UP ROUTINES</strong></td>
<td>Moderator: Maria Barbero</td>
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<td></td>
<td>What should be monitored, when and by whom?</td>
<td>Lena Lande Wekre</td>
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<td></td>
<td>How do we avoid unnecessary complications without</td>
<td>Zoran Radunovic</td>
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<td>causing too much worry?</td>
<td>Inger-Margrethe Paulsen</td>
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<tr>
<td>15.45</td>
<td>Evaluation and closing remarks</td>
<td>Inger-Margrethe Paulsen</td>
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</table>

16.00 Program ends

16.15 Shuttle to Skøyen station (Flytoget Airport Express Train)

People who want to join the shuttle need to register on the list hanging outside the plenary room. There will be no shuttles directly to the airport at this time because of rush hour.
What do we know about adults with osteogenesis imperfecta in Denmark – the non-skeletal issue, eyes and quality of life

Jannie Dahl Hald, MD PhD, Department of Endocrinology, Aarhus University Hospital, Denmark, jahald@rm.dk

Coworkers: Lars Folkestad (Odense University Hospital, Denmark); Torben Harsløf; Christian Heinrich Leonhard; Bente Langdahl (Aarhus University Hospital, Denmark)

Osteogenesis imperfecta (OI) is an inherited disease of the connective tissue with a broad clinical range including skeletal and non-skeletal issues. The aim of the present study was to determine the impact of non-skeletal issues including scleral status and quality of life in a population of adult Danish patients with OI.

A cross-sectional study of 85 Danish adult patients with OI, Sillence type I, III and IV, age 18-78 was conducted. All patients underwent a clinical examination reflecting skeletal and non-skeletal issues including an eye examination by an ophthalmologist. Collagen status was determined by skin fibroblasts, and genetic analyses were performed investigating the COL1A1 and COL1A2 genes, and other genes known to cause recessive OI. Health related quality of life was examined using the SF-36 questionnaire.

The study led to a thorough investigation of adults with OI in Denmark. We found differences in the corneal thickness between the clinically different phenotypes of OI. Mental health related quality of life in this adult population facing many physical challenges was surprisingly similar to an adult Danish population without OI.
What do we know about adults in the Netherlands?

Arian Harsevoort, Msc¹ & Fleur S. van Dijk MD, PhD², Annemarieke Dommissé, MD³, Minka de Jong, Msc⁴, Daniëlle van den Grijn, Msc⁴, Mieke Joostens, MD⁴, Marije van Leeuwen, MD¹, Luuk J. Scheres MD¹, Anton A. Franken MD⁶, PhD, Guus J.M. Janus MD, PhD¹

¹Department of orthopedics, Isala hospital, Zwolle, the Netherlands
²Department of clinical genetics, VU medical center, Amsterdam, the Netherlands
³Department of rehabilitation medicine, Isala hospital, Zwolle, the Netherlands
⁴Department of occupational therapy, Isala hospital, Zwolle, the Netherlands
⁵Department of gynaecology, Isala hospital, Zwolle, the Netherlands
⁶Department of internal medicine, Isala hospital, Zwolle, the Netherlands

The expert center for adult patients with OI was founded in 2007 in the Netherlands (Zwolle, Isala clinics) on specific request of the board of the Dutch OI patient organization (VOI) as they had learned from their members that the needs of adult patients with OI were insufficiently covered by the Dutch health care.

Eight years later, in September 2015, 250 adult patients have visited this expert center. Our first priority has been to deliver up-to-date patient care for patients with OI. However, in order to bring patient care to a higher quality level, we started assembling data from patients with OI in order to explore the natural history of OI and to identify issues that needed to be investigated and addressed in adult patients.

From the first 150 patients we have assembled clinical and genetic data which will be presented. Furthermore, we defined urgent research questions which has led to undertaking of scientific studies exploring among others the following subjects: OI and fatigue, occurrence of atypical femur fractures, intrafamilial variability in OI and OI and pregnancy. We will present the structure and progress of these studies.
Joint laxity and muscle strength in OI – study about children from 0-6 and later consequences in life. Possible impact on activity and participation

*Olga de Vries (physiotherapist), Heidi Johansen (occupational therapist)*  
*Olga.devries@sunnaas.no*

**Introduction**  
Earlier studies of adults with OI showed multiple problems in lower extremities, also in type I. We wanted to explore whether these problems already were present in childhood.

This paper shows the description of joints, muscle function and lower limb axial alignment.

**Methods**  
All children with OI registered at TRS national resource centre aged 0-6 years (N=15) were invited and 12 children (1 – 6 years of age) accepted to participate; seven with OI type I, two with type III, two with type IV and one uncertain type.

These children were followed twice a year for 3 years in a multidisciplinary clinical evaluation.

**Results**  
All children with OI type III and IV had skeletal deformities: bowed long bones, pes planes and scoliosis. Children with OI type I had flexible pes planes, genu valgum and/or increased lordosis. We found joint laxity, stiffness, mal alignment and decreased muscle strength in all types of OI.

Almost all children had reduced scores in some scales on PDMS-2 and M-ABC-2, those with low muscle strength, deformities in tibia and femur expressed the lowest gross motor function. PEDI score showed reduced mobility and CAPE showed low participation in sports activities.

**Discussion**  
To give children with OI good follow up it is necessary to find assessment tools that reveal exactly what the child's problems is caused by. For children with OI motor difficulties can be caused by hypermobile joints, weak muscles or lack of experience. PDMS-2 and M-ABC-2 seems suitable for children with mild forms of OI and these tests confirm that many children with OI struggle with gross motor function and balance. The balance difficulties assumed to be related to muscle strength. Evaluation of muscle strength in the lower extremities is probably important functional outcome measures for children with OI. So, in addition to motor tests it is necessary with good clinical examination and evaluation of strength and their habits of physical activities.

Sunnaas Rehabilitation Hospital- TRS National Resource Centre for Rare Disorders  
NKSD Norwegian Advisory Unit on Rare Disorders
Joint Laxity in Osteogenesis Imperfecta and Skeletal Dysplasias

Michael B. Bober, MD, PhD  
Director, Skeletal Dysplasia Program  
Clinical Genetics  
A.I. duPont Hospital for Children  
Wilmington, DE; USA  
mbober@nemours.org

Collaborators: Tariq Rahman, PhD; Jinyong Wee, MS; Shunji Tomatsu, MD, PhD; Colleen Ditro, DNP, CPNP; Angela Duker, MS, CGC; Rich Kruse, DO; William Mackenzie MD

In an effort to measure the degree of joint laxity in individuals with osteogenesis imperfecta and other skeletal dysplasias, we created a device to measure torque and maximum angle of deflection at the second metacarpophalangeal joint. To date, we have collected data on 89 individuals, aged 4-38 years. The individuals studied included those with: no dysplasia (controls), achondroplasia, Morquio syndrome and OI. Comprehensive data analysis is underway. Preliminary review shows a diagnosis independent relationship between patient age and maximum angle. However, there appears to be differences in the maximum angle between different forms of dysplasia. Further statistical analyses are to be completed and future work will look more closely at correlations between types of joint laxity and function.

Thank You!

Michael B. Bober, M.D., Ph.D  
Director, Skeletal Dysplasia Program  

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Wilmington, DE 19803  
O) 302-651-5916 F) 302-651-5033
The foot in OI

Dr. Ana Mª Bueno Sánchez
Children Section Of The Cot Service

INTRODUCTION
We know that type I collagen can also be found within the ligaments and tendons that conform the locomotor apparatus.

There is a OI, coursing, rather than bone fragility, with hypermobility, muscle weakness and joint instability. These aspects isolated, can become so important that they prevent an independent life. In regards to hyperlaxity, OI patients can be divided into two groups:

- Those who experience it associated to bone fragility, with fractures and variable deformities.
- Those who present scarce bone fragility and variable deformities, but who have a severe, and in many cases disabling hyperlaxity.

The most important pathologies associated to hyperlaxity are as follows:

- Kyphoscoliosis (a pathology that may have multiple causes, but can also be caused by a pathological hyperlaxity)
- A variation in the lower limb axes: genu valgum and genu varus (pathologies that may have multiple causes, but can also be caused by a pathological hyperlaxity)
- Recurring sprains
- Dislocations relapsing of the kneecap
- Basilar invagination? Radial head dislocation?
- Plano-valgus and pronation of the foot

35% of our patients could potentially have a pathological hyperlaxity. These injuries to the ligaments, tendons and bones, along with surgical treatment, can result in extremely severe deformities.

FLAT FEET IN OI
Flat foot in OI, apart from the valgus and pronated foot, has specific characteristics. Patients can develop deformities as a consequence of their walking instability: torsional deformities in the tibias, forefoot rotation, hallux dorsal bunions, hallux valgus and mallet finger. This, in turn, makes it difficult to walk, give an inadequate support and a poor control of balance. Flat feet become painful with time and their treatment is complex: the medical treatment does not improve hyperlaxity as it does in the case of osteoporosis, orthopedic treatment is insufficient and surgical treatment has proven not to be resolutive.

Our experience
Upon revising the cases encountered up until August 2015, we have counted a total of almost 300 surgeries on patients with OI. Only 15 of these were foot surgeries, 9 of which were related to flat foot (3% of the surgeries).

This is due to different reasons. On the one hand, patients reach a point in which they do not want more surgeries than those strictly necessary. On the other hand, certain surgical techniques on feet require immobilization, something that is not recommended in the case of OI.
In addition, developments in individual cases after surgery was analyzed and the results were not positive. Ultimately, we decided to stop recommending this treatment. Surgical treatment we indicated only in special situations.

Conclusions
The best treatment for flat foot associated to OI is physiotherapy and orthopedic treatment:
• To better the walking support by means of orthopedic insoles
• To strengthen the intrinsic foot musculature
• To strengthen the leg musculature

In exceptional cases in which there is an excess of deformity, pain and disability, the surgical treatment may be applied. However, keeping in mind that:
• The type of OI that best responds to surgical treatment is type I.
• During surgical treatment, various techniques must be used simultaneously.
• Surgery should strive for a broadening of the support surface of the ankle bone.
• Tendons should be tautened (especially the posterior tibial tendon)
• Orthotics and physiotherapy must follow subsequent to the operation.
Alterations of the lower limb axes

Dr. Ana Mª Bueno Sánchez
Children Section Of The Cot Service

The load bearing axis and the rotational of the lower limbs can be affected due to many causes: the anatomical features of each patient, the inclination of the pelvis, the axis of the load and angle of the hip (varus/valgus, anteversion/retroversion), the articular surface of the knee, the ankle or foot, etc. The fractures previous and deformities previous of the patients, the surgeries performed on the long bones of the lower limbs as well as the special hyperlaxity and debility of soft tissue of each patient are also important factors to be taken into account.

The deformity is sometimes so significant that it is impossible to determine how it reached its present state.

The important issue is to prevent this state of deformity.

How to prevent deformities

1. Physiotherapy. The physiotherapy generally does not have the capacity to improve the tone of ligaments that stabilize articulations, however, this can to improve the muscle's tone. Improve the muscle tone and learning can help control the support and rotatory of the lower extremities.

2. The use of or articulatory orthetic stabilizers. The orthetics are not very popular and they may in many occasions be difficult to adapt to each patient: the size of the thigh, the associated deformities, etc. They must be used in association with physiotherapy because they can cause muscle atrophy.


Surgical treatment in deformities of the lower limbs

When carried out due to fractures or deformities, surgical treatment must aim to never alter the load bearing axis or aggravate articulatory rotation. In fact, during surgery, must be corrected any defect present of the axis or the rotational.

When planning a surgical intervention of the lower limbs should be taken into consideration how the spine, hips, etc.

If the surgical intervention takes place in the later years of infancy, any dysmetry should be accounted for and compensated.

Deformities that are a consequence of hyperlaxity of the knees and ankles can be corrected in children with percutaneous surgery before the closure of growth cartilage, and redirecting the remaining growth. This percutaneous surgery is also used on patients without OI, but must be adapted to the characteristics of the OI bone.
Conclusions
The essential issue is to prevent severe deformities.

In order to reach this goal, the doctor, the physiotherapist, the orthopedic surgeon and THE PATIENT must work together and cooperate as a team.

Although it is the traumatologist who recommends surgery of the growth cartilage tissue, it is you, the patients, who decide if you want to go through with it.

Surgical treatment of deformities in adults is more complex and can lead to difficulties and complications. This type of surgery on adults should be valued by the entire team, and in this case it is the patient who is most important when reaching the final decision. The patient must think about the genuine necessity of the surgical intervention, if it will be as efficient as he wishes and if there are means to work to the post-surgical recovery.

It is very important to think the physical and psychological fatigue of the patient.

At the medical consultation, the patient and his surgeon should take into consideration all of these factors, which ultimately add up to one bottom line: the patient, who is risking, what benefits you can get and how much it will cost to achieve them.
Bone – Muscle unit and Neuromuscular Function in relation to Mobility

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The main function of bone is to supply mechanical support for locomotion. The largest forces that act on bone are caused by muscle contractions. According to Harold Frost’s mechanostat model bone strength is mainly adapted to the voluntary muscle force to avoid bone damage by muscle contractions. As an example, at the lower leg due to the lever ratios forces may act on the tibia that exceed the body weight more than tenfold. These forces cause a deformation of bone. If this deformation exceeds a certain threshold bone strength is increased. On the other hand if a lower threshold is not regularly exceeded bone mass is reduced. This optimization process forms a bone that is adapted to magnitude and direction of muscle forces with minimal bone mass. The sensitivity of bone cells to deformation can be modulated by different factors like hormones, nutrition or genetic factors. At the same time a loss of muscle force reduces bone strength and increases fall risk and therefore increases the fracture risk.

Muscle function can be mainly described by three different parameters: Force, movement velocity and power. While force in relation to body weight is relatively constant over age, muscle power in relation to body weight increases during growth up to the age of 20 – 30 years and declines afterwards permanently with age. Power can be generated at low levels over long periods of time (endurance) or at high levels for short periods (chair rising, jumping). For mobility the high power over short time is more important than endurance. Mechanography is a suitable tool to evaluate muscle function.

In general the muscle can work in three different functions: to generate energy (e.g. cycling, chair rising), to store energy (e.g. walking, hopping) or to dissipate energy (shock absorber, walk down stairs).

Different training regimens are available to improve the different aspect of muscle function and may therefore help to maintain mobility during ageing and disease.
Rib cage deformities alter respiratory muscle action and chest wall function in patients with severe OI

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**Background.** Osteogenesis imperfecta (OI) is an inherited connective tissue disorder characterized by bone fragility, multiple fractures and significant chest wall deformities. Cardiopulmonary insufficiency is the leading cause of death in these patients.

**Methods.** Seven patients with severe OI type III, 15 with moderate OI type IV and 26 healthy subjects were studied. In addition to standard spirometry, rib cage geometry, breathing pattern and regional chest wall volume changes at rest in seated and supine position were assessed by opto-electronic plethysmography to investigate if structural modifications of the rib cage in OI have consequences on ventilatory pattern. One-way or two-way analysis of variance was performed to compare the results between the three groups and the two postures.

**Results.** Both OI type III and IV patients showed reduced FVC and FEV1 compared to predicted values, on condition that updated reference equations are considered. In both positions, ventilation was lower in OI patients than control because of lower tidal volume (p<0.01). In contrast to OI type IV patients, whose chest wall geometry and function was normal, OI type III patients were characterized by reduced (p<0.01) angle at the sternum (pectus carinatum), paradoxical inspiratory inward motion of the pulmonary rib cage, significant thoraco-abdominal asynchronies and rib cage distortions in supine position (p<0.001).

**Conclusions.** In conclusion, the restrictive respiratory pattern of Osteogenesis Imperfecta is closely related to the severity of the disease and to the sternal deformities. Pectus carinatum characterizes OI type III patients and alters respiratory muscles coordination, leading to chest wall and rib cage distortions and an inefficient ventilator pattern. OI type IV form is characterized by lower alterations in the respiratory function. These findings suggest that functional assessment and treatment of OI should be differentiated in these two forms of the disease.
Causes of death in patients with Osteogenesis Imperfecta, a registry based trial

M.D Lars Folkestad, PHD student, Odense University hospital Department of Endocrinology and University of Southern Denmark Institute of Clinical Research.

Osteogenesis Imperfecta (OI) is a hereditary disease caused by mutations to the genes that facilitate the biosynthesis of collagen type 1. The most prevalent symptom of OI is increased fracture-risk and -rate and varying degrees of bone deformities and growth retardation. Where the bone phenotype of OI is well described, less is known about the other manifestations of the disease. In Denmark causes of death and the time of death have been registered since 1875. All hospital admissions, from 1977, and out-patient-clinic contacts, from 1995, are registered via the National patient registry (NPR). All citizens are given, from birth or from date of residency in Denmark, a unique Central Persons Registry (CPR) number enabling us to connect the data from different registries on an individual level. This makes it ideal to investigate the causes of death in the Danish OI population. The WHO ICD-8 classification was used until 1995 where the ICD-10 classification was introduced.

According to a study, from 1996, McAllison and Paterson* found that the most frequent cause of death in OI was lower respiratory tract infections. We hypothesise that patients with OI in Denmark have an increased risk of death caused by respiratory tract infections, and aim to investigate the most common causes of death in a historic cohort study and comparing the OI population to healthy (non-OI-diagnosed) controls.

Using the Danish health registries, we identified all patients with either an ICD-8 or ICD-10 OI diagnosis in the National Patient Registry (NPR), using the Danish Civil Registry System and Central person number (CPR) we matched every patient to 5 controls. We matched the controls on age, gender and excluded persons in first- and second-degree relatives to a case from being a control. Cases could not also be controls and controls could not become cases. For all participants data regarding date of birth, date of death, manner of death, place of death and the underlying and contributory causes of death. Data is presented descriptively.

We found that the most common cause of death was found to be OI, cardiovascular disease and malignancy, compared to the control group where malignancy was the leading cause of death, followed by cardiovascular disease. None of the patients died due to an infection. All but 8 of the 112 recorded deaths in the OI cohort died of natural causes.

The data is based on registry data. Using registry you are limited to and limited by the conclusions and actions of the physicians who see the patients. We cannot rule out that some of the patients identified have been mis-classified. Furthermore we cannot rule out that not all persons with OI have been in contact with an outpatient clinic or admitted to a hospital – thus not showing up in the registries. No knowledge of OI subtype, genetic disposition can be drawn from the data, which is a major limitation to our data. One of the strengths of the study is that the study is population based and including all known cases of OI.

In conclusion, we found using the Danish health registries that the most prevalent cause of death in the Danish OI cohort was OI, followed by cardiovascular disease and malignancy. Further investigation into the contributory causes of death and evaluation of confounders such as competitive risk is needed.

Chronic pain and OI

Tamara Fernández, Psychologist
AHUCE – Asociación Nacional Huesos de Cristal OI España

Osteogenesis imperfecta (OI) comprises a heterogeneous group of genetic disorders characterized by increased bone fragility, low bone mass, and susceptibility to bone fractures with variable severity. There are some clinical signs that can appear in people with OI: bone deformities, blue sclerae, hearing loss, dentinogenesis imperfecta and chronic pain.

The International Association for the Study of Pain (IASP) defines pain as an unpleasant sensory and emotional experience associated with actual or potential tissue damage, or described in terms of such damage. According to the duration of the pain it can be classified in acute and chronic pain. Acute pain begins suddenly and is usually sharp in quality. It serves as a warning of disease or a threat to the body. It might be mild and last just a moment. In most cases it does not last longer than six months, and it disappears when the underlying cause of pain has been treated or has healed. Chronic pain persists despite the fact that the injury has healed (more than six months). Pain signals remain active in the nervous system for weeks, months, or years. Chronic pain produces physical effects (tense muscles, limited mobility, a lack of energy and changes in appetite) and emotional effects (depression, anger, anxiety, and fear of re-injury). It might have originated with an initial trauma/injury or infection, or there might be an ongoing cause of pain.

In order to analyse what is the chronic pain prevalence in OI in Spain, from AHUCE’s physiotherapy and psychology departments, we have started a research study about it. First descriptive results show that a 52% of the people suffer pain nowadays, and a 36% of them are suffering pain for more than six months. We are currently working on this study and we are wondering to show all the results soon.

Treating pain in OI: Although medical treatment plays a major role in the management of OI, psychological strategies are critical components of this plan. Psychological therapies result an improvement in pain relief across several different pain conditions. Although most individuals with OI receive adequate medical care, most do not have access to basic education about behavioral and cognitive strategies, including pain management, which has been shown to make a difference to the lives of other people with chronic pain.

Pain Less - OI-youngsters take control

This is the reason for running an international proposal to manage chronic pain: Pain less – OI youngsters take control. This project aims to develop an online pain-management program for young people (14-27 years old) with OI. The program integrates evidence-based methodologies of pain self-management, behavioral and cognitive interventions; also incorporating sections from two existing online, empirically validated programs, directed to adolescents with chronic pain conditions, but adapting its contents to the specificities of OI.

At the moment we are in the program translation and adaptation phase.
Fatigue in adults with OI and other connective tissue disorders

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Is fatigue an issue for adults with OI? In the clinic we meet some adults who describe the experience of fatigue as a limitation in their daily lives. As to date few studies explore both prevalence and associations to fatigue in this population. Knowledge on fatigue in other connective tissue disorders like Ehlers-Danlos syndrome and Marfan syndrome might be applicable also in OI, both concerning results and methodology.

The presentation will give a short explanation on the concept of fatigue, then a short overview on available studies on fatigue in Ehlers–Danlos syndrome.

Results from a cross-sectional study on prevalence of fatigue and associated factors in Marfan syndrome will be presented. Main findings were that prevalence of severe fatigue was higher in adults with Marfan syndrome than in the general population. Fatigue was significantly associated to chronic pain and employment status.

A literature search on fatigue, vitality and chronic pain in adults with OI will be presented, aiming to discuss what is known about prevalence and possible associations to fatigue in adults with OI.

Concludingly draw up questions on need for further research on fatigue in OI.
Coping strategies in adults with OI

Dr. Alessandra Ciliberti  
Psychologist Psychotherapist, As.It.O.I., Italy

Data about pain in OI Adult patients are a part of a larger Italian pilot study: “Psychological and social aspects in OI”. A two-years (April 2013-2015) pilot study supported by As.It.O.I. and realized at “La Nostra Famiglia” Institute in Bosisio Parini (Lecco, Italy).

The study focused on:

- Coping strategies adopted by OIer’s to manage the internal and external demands of situations that are appraised as stressful. The way patients and families cope with illness/disability related stress has been associated with health outcomes, quality of life, adherence and psychosocial adjustment.
- Perceived quality of life by people with OI.
- OI’s personality structural indexes (as Sel af Perception, Interpersonal Relation, Affects).

About pain: about 56% of our OI adult sample declares to suffer from pain (77% of women and 28% of men).

We consider statistically significant data (Chi square test, p<0,05): people who suffer from pain feel to be exposed to situational distress, demonstrate lower rating in positive attitude coping style, are dissatisfied with their health, are less able to get around, judge low their quality of life in physical area, are not so aware of their inner emotions and thoughts and are less capable to express them in psychological terms, need and reach out for physical and emotional closeness.

After presenting some neurobiological data about different kind of pain we stress the importance to work together with the patient on his prevalent coping style for pain to evidence maladaptive pattern, negative memories regarding pain, avoidance coping style that makes impossible to accept medical conditions. As Psychotherapist our target is working with patient to re-connect sensation, emotions and feeling in order to install new coping strategies, enforce personal resources, giving a new perception of pain experience and a new sense of control on it.

Useful tools to facilitate verbal communication on pain both with adults and children are discussed.
Perfectly normal – or The Odd One Out?

Elise Christensen  
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**Institute/organization I work for:** TRS National Resource Centre for Rare Disorders, Sunnaas Rehabilitation Hospital, Nesoddtangen, Norway  
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**Abstract:**  
In a qualitative study conducted at TRS Resource Centre for Rare Disorders the experience of living with a physical disability in social settings was explored. In total 8 adults with AMC participated in focus group interviews. In group 1 the participants were ambulants, while in group 2 the participants were wheelchair users. The findings showed that the degree of disability does not necessarily determine how the participants look at themselves. The participants, who were ambulants, expressed more fatigue and concerns about being different than the participants who were wheelchair users. Theories about stigma may help explain why this might be the case, as this could be a consequence of the process of stigmatization.

The study also highlights that in some ways it can possibly be more challenging to live with a less severe than a more severe disability.

The findings from this study will be related to adults living with OI. How might it be experienced living with a mild and invisible type of OI compared with living with a more severe and more visible type of OI? In which ways might this also influence their experience of fatigue?
Exercise as pain relief for adults with OI

Ann Bett Kirkebæk, Denmark
Certified Pilates- and Fitness instructor, lecturer, coach and personal trainer.
CEO and founder: Pilates in Aarhus by Ann Bett Kirkebæk

Short on research by Danish Ph.D and physiotherapist Henrik Bjarke Vægter.

The research has proved how both low as well as high intensity exercise gives pain relief immediately and even long lasting pain relief. In fact, exercise can give you the benefit of pain relief on an equal level as resting in bed.

The research also indicated that parts of the body that are not doing exercise will benefit from parts of the body that are doing exercise.

Short on research; The ATPBone project, led by danish researcher and professor, Ph.D, dr. med. Niklas Rye Jørgensen.

The ATPBone project was mainly just focused on the more basic mechanisms for the regulation of bone turnover and in vivo experiments to explore specific P2 receptors involved in this regulation. The project proved that physical exercise with weight bearing activities releases ATP which helps maintaining bone cells and accelerates the healing of broken bones.

The ATPBone project also indicated that ATP release and P2 receptors are involved in pain regulation too. Therefore we can assume that physical exercise is preferable not only to achieve stronger bones but also achieving pain relief.

Main conclusions:
We know from research that the whole body will benefit from exercises only performed by/on isolated parts of the body. Not only to achieve stronger bones but also achieving pain relief. Therefore we can assume that (isometric) exercises on the upper body also will bring pain relief to the lower body. This knowledge is vital for adults with OI – especially those who use wheelchair.

Pain relief will bring the body more energy for exercise which will in turn cause additional pain relief that gives renewed energy for doing exercise.
The Brittle Bone Disorders Rare Disease Clinical Research Consortium (and contact registry)

Ingunn Westerheim  
Chairperson of NFOI & President-Elect of the OIFE

During the 7th international conference in OI in Salzburg, the OIFE and NFOI had a meeting with Tracy Heart from the OIF, dr. Brendan Lee, dr. Glorieux and dr. Marini. One of the topics was how we can spread information about the different research projects going on and how we can collaborate on registries. The following information is gathered from www.oif.org and www.rarediseasesnetwork.org/cms/BBD

The Brittle Bone Disorders Consortium is part of the National Institutes of Health Rare Diseases Clinical Research Network in America. It is a multi-center program that focuses on understanding and providing better treatment options for all types of OI. In addition, the consortium will sponsor a Contact Registry for People with OI and implement training programs for physicians and scientists.

What this Means to People with OI
This program will speed up the pace of research and put useful information into practice much quicker than otherwise possible by:

- Using the skills of experienced researchers from many institutions
- Creating access to research centers closer to where people live; making participation easier
- Expanding educational opportunities for healthcare providers

Consortium Members
The Brittle Bone Disorders Consortium is a group of physicians, researchers and educators who are focused on learning more about OI and developing new and better treatments to improve the care of people with OI.

- Baylor College of Medicine, Houston, TX
- Shriners Hospital for Children Montreal, Canada
- Oregon Health and Science University, Portland, OR
- University of California in Los Angeles
- Kennedy Krieger Institute, Baltimore, MD
- Children’s National Medical Center, Washington, DC
- Hospital for Special Surgery, New York, NY
- Shriners Hospital for Children, Chicago, IL and Marquette University, Milwaukee, WI
- The University of South Florida, Tampa, FL
- University of Washington, Seattle, WA
- OI Foundation, Gaithersburg, MD
Contact Registry for People with OI
This Consortium project is a state of the art registry of contact information for children and adults who have OI. It makes it possible to do the following:

- Expand knowledge about OI through the information provided by registry members.
- Reach out quickly to people with OI who are eligible for a new study
- Update registry members on study results and other news.

Patients with OI living in any country are encouraged to join the Contact Registry in the US. To join the Contact Registry, go to www.rarediseasesnetwork.org/cms/BBD

Consortium Research Studies
Extending the Longitudinal Study of OI (begun in the OI Foundation’s Linked Clinical Research Center program). This study is important to accurately describe the different types of OI and how they may change and as people grow up and get older. This study is recruiting participants - see Natural History Study (or Longitudinal Study) under Current Studies.

Medical Education Opportunities and Grants
The Brittle Bone Disorders Consortium, with the assistance of the OI Foundation, will work to bring up-to-date information about caring for people with OI to physicians and other health care providers. The OI Foundation also has grants for investigators. See www.oif.org.

Care4BrittleBones
Another source investigators who work with OI should know about is Care4BrittleBones. The Care4BrittleBones is an international charity with roots in the Netherlands. It was founded in 2012 by family and friends of a child with OI, and the foundation has grown to become the biggest fundraising organization for OI research in Europe. Care4BB are recognized as a charitable organization in the Netherlands, which allows for tax deductability of donations across the entire EU. Even if most of the funds that have been raised until now comes from the Netherlands, the foundation has opened for applications from investigators from all over the world. The purpose of this is to spend the money where they can be most efficiently used.

See www.care4brittlebones.org
The RUDY-study – a novel approach to patient driven research

Dr Lydia Forestier-Zhang  
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There are currently 456 rare bone diseases recorded within 40 groups. Within many of these diagnoses there is marked heterogeneity of severity and complications that is often not explained by current understanding of disease mechanisms. There is an urgent need to improve the care of these patients by developing novel diagnostic tests and therapies based on understanding sub-phenotypes within existing diagnostic groups.

The rare diseases of bones, joints and vessels (RUDY) study aims to develop a national cohort of participants with rare disorders of bones, joints or blood vessels in the UK from which to increase understanding of disease mechanisms for sub-phenotyping.

Participants aged 0-100 years with clinical diagnosis of a rare disorder of bones, joints or blood vessels are recruited via the study website www.rudystudy.org. Participants complete online questionnaires for health-related quality of life, pain and function questionnaires every 6 months. Further phenotyping including physical examinations, DXA scans and blood and urine tests are performed depending on individual projects.

The secure open-source RUDY database is now online and includes features such as web based registration, dynamic consent and online assessment and two-way communication. These features have been tested by over 300 to date participants successfully with good feedback. Further, this has brought together specialist centres across the UK from both paediatric and adult services within a single clinical and academic network.

The RUDY database has delivered a novel approach to recruit and standardize assessment patients across the life-course with rare diseases. The platform offers opportunities for international collaboration and cohort based research.
Adult clinic in Birmingham – West Midlands Adult OI Service – Need and Service Provision

Susan Stewart
Endocrine Genetic Clinical Nurse Specialist (Birmingham Women’s Hospital and Queen Elizabeth Hospital Birmingham)

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OI Bone Clinic Colleagues
Dr Neil Gittoes,
Consultant Metabolic Bone Specialist, Queen Elizabeth Hospital Birmingham
Dr Trevor Cole
Consultant Clinical Geneticist, West Midlands Clinical Genetics Unit, Birmingham Women’s Hospital

In the UK, the West Midlands offers one of four recognised complex childhood Osteogenesis Imperfecta (OI) services (NHS England), and has close links to the regional genetic service. Until recently, on reaching adulthood, more severely affected young adults remained under the care of the paediatric service, while patients with milder phenotypes were discharged to back to routine primary care.

In 2007, an audit of OI-patients known to the clinical genetics service revealed a large cohort (254) of patients and potentially affected family members unknown to our regional adult metabolic bone service. It is well recognised that older adults with OI are at greater risk of fracturing than the general population and therefore a significant number may benefit from a specialist adult service. Further contact with our audit patient cohort then initiated discussion of clinical need between adult service providers.

Our adult OI clinic commenced in 2008 and sits within the metabolic bone/ genetic service. It is now well established and includes adult patients of all ages, from across the UK.

Many adults with OI require ongoing support, even though not actively fracturing, and our clinic’s aim is to coordinate holistic care on a needs basis and provide a point of contact when further difficulties arise. We have links with many other clinical hospital services as required, and we also liaise closely with the patient’s community practitioners.

Our regular clinical team consists a metabolic bone specialist and consultant clinical geneticist, both with specialist interest in OI, and an endocrine/genetic clinical nurse specialist (point of contact and liaison)

OI is a highly individual condition with family-wide consequences and best practise requires a multidisciplinary team approach. Nurses are in a unique position to facilitate the partnership between care-providers, patient and family. (Steiner RD and Adsit J et al, 2005, updated 2014)
Adult care in the Netherlands

Arian Harsevoort, Msc\(^1\), Annemarieke Dommisse, MD\(^2\), Fleur S. van Dijk, MD, PhD\(^3\), Minka de Jong, Msc\(^4\), Daniëlle van den Grijn, Msc\(^4\), Anton A. Franken MD, PhD\(^5\), Guus J.M. Janus, MD, PhD\(^6\)

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The expert center for adult patients with OI was founded in 2007 in the Netherlands (Zwolle, Isala clinics) on specific request of the board of the Dutch OI patient organization (VOI) as they had learned from their members that the needs of adult patients with OI were insufficiently covered by the Dutch health care.

Eight years later, in September 2015, 250 adult patients have visited this expert center. Our first priority has been to deliver up-to-date patient care for patients with OI. The multidisciplinary approach and experience will be discussed.
What do we want?

Ingunn Westerheim  
Chairperson of NFOI & President-Elect OIFE

In my talk I will raise some questions for the debate about adult services. But I start off with the challenges of having a rare disease like OI:

◦ Little knowledge and awareness
◦ The dilemma of “all or nothing” (everything is blamed on the OI; or you forget the OI when you are treating a common disease)
◦ A need for services throughout the lifespan
◦ Going from one phase in life to another (transition) can often create challenges
◦ Need for interdisciplinary and coordinated services
◦ “No straight production lines” - complex challenges

When you are a child – you receive services from the children’s hospital where the pediatrician gives you treatment and regular follow-up. As an adult you are regarded as a collection of body parts that can have their own complications. And these body parts belong in separate houses with different professionals having different levels of knowledge.

Who should coordinate? It’s not possible to get enough knowledge for each rare diagnose in the local communities. So we need access to specialized services - evaluation and treatment from someone who knows OI. As a minimum some treatments should be given regionally and perhaps even nationally (or internationally). We also need access to regular follow-up to prevent complications.

Maybe an “OI-house” is the answer – where you can, not only receive advice, but also:

◦ A diagnose
◦ Treatment
◦ Follow-up
◦ Be a part of research projects – which leads to new knowledge

The “OI-house” is not the same as the Norwegian version of the Centre of Expertise (TRS). TRS is supposed to build and spread knowledge about OI. They also have individual consultations and know a lot about OI. But TRS don’t provide treatment and regular follow-up and they are not tightly connected to a clinic. We want the best of both worlds!

The dilemma is that not only do we want a doctor who knows OI, but we want the best cardiologist and the best eye doctor. How do we solve this?

And who should work in the OI-house? And how do they collaborate with the Centre of Expertise? And how do they collaborate with the best ear doctors, the best underarm surgeons and the best cardiologists? Do we need an expert co-ordinator instead?
Follow-up routines of adults with OI – Does age make any difference to the need of care?

Lena Lande Wekre, MD, Phd
Norwegian National Advisory Unit on Rare Disorders, NKSD, Oslo University Hospital

Osteogenesis imperfecta (OI) is a genetic disorder of increased bone fragility and other connective-tissue manifestations with a wide spectrum of clinical expressions. Even though the main problems come from the skeleton, we have to make an overall examination including skeletal deformities, joints and muscles, hearing, sight, teeth, heart, lungs and gastrointestinal organs to establish optimal follow up guidelines for people with OI. These examinations should then provide the basis for an individually adapted follow up where one has taken into account type of OI, age and specific findings.

There are several studies and reports on follow up routines and treatment of children with OI. Few studies have however described clinical and social aspects in adults with OI, and especially the consequences of ageing. Distinctions may be made between the biologic ageing process (age changes that all people share), and the “probabilistic ageing process” (age changes that may happen to some, but not all people as they grow older) which may be influenced by OI. This overlap of processes should also be taken into consideration when making follow up programs for adults.

The plan for supervision is meant to give information needed to prevent complications related to OI, and to limit the loss of function. It should be a tool for persons with OI (and their families), the GPs and other health-professionals who are giving care to persons with OI.

We suggest that regular follow up of adults with OI, customized clinical findings and function of each individual, should be conducted in Regional Hospitals in collaboration with the local doctors (GPs)

The presentation will give a systematic overview over key clinical alterations and suggestions for follow up routines.
Follow-up of heart issues

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All registered adults with OI in Norway above the age of 25, were invited to participate in the Norwegian survey of adults with OI.

We studied 99 subjects, 58 women and 42 men who agreed to participate, at the Cardiology Department, Oslo University Hospital (Aker), from June 2003 to October 2007. The 52 recruited control subjects were sex- and age-matched and did not have history of cardiovascular disease, hypertension, diabetes, or use of cardiac medications.

Patients were divided into type I, III, and IV, and 52 control subjects. History and physical examination, ECG, and echocardiographic parameters of left ventricular (LV) and right ventricular (RV) systolic and diastolic function were obtained.