

# Scientific Programme

August 27<sup>th</sup> - 30<sup>th</sup> 2017

ALL SESSIONS ARE IN **EXPO HALL** UNLESS OTHERWISE INDICATED

## Programme: Sunday August 27th

**17:00 - 17.20**

### Welcome address

**Chairs:** Lena Lande Wekre (Programme Chair)  
Inger-Margrethe Stavdal Paulsen (Chair of NFOI)

**17:20 - 18.45**

### Keynote lectures

**17.20 - 18.00**

**Biomimicry – could it help us find novel treatment strategies for human disease?**

Peter Stenvinkel (Stockholm, Sweden)

**18.00 - 18.40**

**The Role of the Osteocyte in Muscle/Bone Crosstalk**

Lynda Bonewald (Kansas City, US)

**18.45 - 20.00**

### Welcome Reception

EXPO HALL FOYER

*Followed by free time*

## Programme: Monday August 28th

**09:00 - 09:15** Opening remarks

**09:15 - 11:00**

**Session 1** Genotype - Phenotype - Diagnosis

Moderator: David Sillence (Sydney, Australia)

**Invited speaker: Paul Coucke (Ghent, Belgium):**

Further insights in the pathogenetic mechanism of OI through human and zebrafish

## Oral Communications

**O1**

**Growth Characteristics in Osteogenesis Imperfecta – Results from an Observational Study from the Linked Clinical Research Centers and the Brittle Bone Disorders Consortium**

Vernon Reid Sutton, Houston, US

**O2**

**Longitudinal Growth Curves for OI Caused by Structural Mutations in Type I Collagen**

Joan Marini, Bethesda, MD, US

**O3**

**A familial FKBP10 mutation associated with a wide phenotypic spectrum ranging from Arthrogryposis to OI**

Osama Essawi, Ghent, Belgium

**O4**

**Use of patient-specific iPSCs as a platform for genotype-phenotype analysis of OI**

Xiaonan Xin, University of Connecticut, US

**O5**

**SPARC-related osteogenesis imperfecta with a myopathy-like presentation**

Cecilie Rustad, Oslo, Norway

**11.00 - 11.30**

**Coffee break**

**11:30 - 13:00**

**Session 2 Collagen: mutations and altered modification**

Moderator: Joan Marini (Bethesda, US)

**Invited speaker: Ruud A Bank (Groningen, The Netherlands)**

Molecular insights into lysyl hydroxylation and cross-linking of fibrillar collagens in normal and diseased bone

**Invited speaker: Richa Garva (Manchester, UK)**

The clock ticks for collagen

## Oral Communications

**O6**

**Biochemical characterization of a COL1A1 signal peptide heterozygous mutation leading to severe OI**

Uschi Lindert, Zurich, Switzerland

**O7**

**Chemical chaperone treatment ameliorates cellular homeostasis of patients with Osteogenesis Imperfecta**

Antonella Forlino, Pavia, Italy

**13:00 - 14:00**

**Buffet lunch/Exhibits/Posters**

*Lunch is served in Brasserie X, the hotel restaurant.*

**14:00 - 15:00**

**Session 3**

**Debate: "This house believes that stem cell transplant is the optimal treatment of OI"**

Moderator: Bente Lomholt Langdahl (Århus, Denmark)

**Invited speakers:** Cecilia Götherström (Stockholm, Sweden)  
Nick Bishop (Sheffield, UK)

**15.00 - 15.15 Coffee break**

**15:15 – 17:00**

**Session 4 Orthopaedic Developments**

Moderator: Richard Kruse (Wilmington, DE, US)

**Invited speaker: Thomas Wirth (Stuttgart, Germany)**

Surgery in upper extremities in OI

**Oral communications**

**O8**

**Incidence and treatment of femur fractures in adults with Osteogenesis Imperfecta**

Alexander Goudriaan, Zwolle, The Netherlands

**O9**

**Challenges of treating the tibia in patients with osteogenesis imperfecta. A single surgeon series**

Darko Anticevic, Zagreb, Croatia

**O10**

**Results of combined intramedullary nailing and supplemental fixation of long bones in patients with osteogenesis imperfecta (OI)**

Matthias Rogalski, Berlin, Germany

**O11**

**Management of the Upper Extremity in OI: 20+ years' experience**

Kathleen Montpetit & Francois Fassier, Montreal, Canada

**O12**

**Treatment of Scoliosis in Osteogenesis Imperfecta with Cement-augmented Pedicle Screw Instrumentation: A Preliminary Experience in Single Institute**

Richard Kruse, Wilmington, DE, US

**O13**

**Acetabular Protrusio in Osteogenesis Imperfecta: Progression and Risk Factors**

Junho Ahn, New York, US

**O14**

**Prevalence and progression of spondylistheses in children with Osteogenesis Imperfecta**

Verity Pacey, Macquarie University, Australia

**17.30**

**Social programme (faculty dinner & sightseeing tour for participants)**

## Programme: Tuesday August 29th

**09:00 - 09:15** Opening remarks

**09:15 - 11:00**

### Session 5 The Connective Tissue Spectrum in OI

Moderator: Jannie Dahl Hald (Århus, Denmark)

#### Invited speakers:

##### Lars Folkestad (Odense, Denmark)

Cardiovascular diseases in patients with Osteogenesis Imperfecta – a nationwide, register-based cohort study

##### Barbro Malmgren (Stockholm, Sweden)

Tooth agenesis in osteogenesis imperfecta; prevalence, mutations and therapeutic considerations

##### Hollis Chaney (Washington, US)

Pulmonary Complications of Osteogenesis Imperfecta

### Oral Communications

**O15**

#### Scoliosis and Cardiopulmonary Outcomes in Osteogenesis Imperfecta

Cathleen Raggio, New York, US

**O17**

#### Inflammation and increased osteoclastogenesis in osteogenesis imperfecta murine

Ivo Kalajzic, Farmington

**O16**

#### Dental occlusion and temporomandibular disorders in adults with osteogenesis imperfecta

Hans Gjørup, Aarhus, Denmark

**11.00 - 11.30**

**Coffee break**

**11:30 - 13:00**

### Session 6 Bone Tissue in Human OI, Mice and Z-fish models

Moderator: Peter Byers (Seattle, US)

#### Invited speaker: Nadja Fratzi-Zelman (Vienna, Austria)

Bone material characteristics in human Osteogenesis imperfecta (OI) and mouse models

### Oral communications

**O18**

#### Collagen C-propeptide Cleavage Deficiency Increases Bone Mineralization and Alters Bone Cell Differentiation

Aileen Barnes, Bethesda, MD, US

**O19**

#### Altered Bone Nano- and Microstructure in Osteogenesis Imperfecta caused by a C-propeptide Cleavage site Mutation

Jannie Dahl Hald, Aarhus, Denmark

**O20**

**Murine model for type VI OI (Serpinf1-/-) reveals dynamic regulation of vascularization and mineralization in bone**

Heeseog Kang, Bethesda, MD, US

**O22**

**Generation of recessive osteogenesis imperfecta zebrafish models using CRISPR-Cas9 system**

Francesca Tonelli, Pavia, Italy

**O21**

**Zebrafish type I collagen mutants show a spectrum of skeletal phenotypes mimicking the clinical variability in OI**

Andy Willaert, Ghent, Belgium

**13:00 - 14.00**

**Buffet lunch/Exhibits/Posters**

*Lunch is served in Brasserie X, the hotel restaurant.*

**14:00 - 15:25**

**Session 7      Current status in treatment - Expo Hall 2 + 3**

Moderator: Francis Glorieux (Montreal, Canada)

**Invited speaker: Brendan Lee (Texas, US)**

Implications of Excessive TGF $\beta$  signaling in Collagen-related forms of OI on Treatment

### Oral communications

**O23**

**Combined effects of sclerostin antibody and pamidronate induce distinct advantages for OI treatment in Brl/+ mouse**

Ken Kozloff, Ann Arbor

**O26**

**Characterization of Chihuahua as a dominant osteogenesis imperfecta zebrafish model and its use in new pharmacological approaches**

Antonella Forlino, Pavia, Italy

**O24**

**Cathepsin K knockout reduces fracture incidence and improves trabecular bone mass and microarchitecture in the axial skeleton of oim/oim mice**

Thomas Roels, Brussels, Belgium

**O27**

**Use of induced pluripotent stem cells and CRISPR/Cas9 in Osteogenesis Imperfecta: first results**

Fleur van Dijk, Groningen, The Netherlands

**O25**

**Screening for osteogenic compounds using the zebrafish as a model**

Jan Willem Bek, Ghent, Belgium

**15.25 - 15.45**

**Coffee break**

**15:45 - 17:05**

**Parallell workshops**

**WS1. Unmet needs in preclinical/animal research (basic) - *Idérommet***

Moderator: Oliver Semler (Cologne, Germany)

**Introduction to discussion: Chris Niyibizi (Hershey, US):**

Animal models of osteogenesis imperfecta and their application in testing novel therapies

**Debate**

**WS2. Follow up in OI – all ages (clinical) - *Expo Hall 2 + 3***

Moderator: Eva Åström (Stockholm, Sweden)

**Introduction to discussion: Francis Glorieux** (Montreal, Canada) – children

**Lars Folkestad** (Denmark) / **Lena L. Wekre** (Norway) – adults

**Debate**

**17.05 - 17.30**

**Coffee break**

**17:30 - 18:30**

**Session 8 Rehabilitation /Pain /Quality of life (in parallell with Workshop - Orthopaedic techniques) - *Expo Hall 2 + 3***

Moderator: Lena Lande Wekre (Oslo, Norway)

**Oral communications**

**O28**

**Physical activity levels and perceived barriers to participation of children with Osteogenesis Imperfecta**

Verity Pacey, Macquarie University, Australia

**O31**

**Preventative Care and Treatment in Osteogenesis Imperfecta: What role for Clinical Psychology?**

Megan Riddington & Rebecca Jones, London, UK

**O30**

**Osteogenesis Imperfecta Foundation: New Strategies in Advancing Education of Individuals, Families and Healthcare Providers**

Tracy Hart, Gaithersburg, US

**17:30 - 18:45**

**WS3 - Orthopaedic techniques (in parallell with session 8) – *Idérommet***

Moderators: Ivan Hvid (Oslo, Norway) & Thomas Wirth (Stuttgart, Germany)

**Introductions to discussion:**

Francois Fassier (Montreal, Canada)

Miguel Galban (Medellín, Colombia)

Darko Anticevic (Zagreb, Croatia)

Inger Holm (Oslo, Norway)

**20:00 - 23:00 Aperitif & Conference dinner**

*Aperitif is served in the Reception Bar. Conference dinner is served in Expo Hall.*

## Programme: **Wednesday August 30th**

**09:00 - 09:15** Opening remarks

**09:15 - 10:45**

**Session 9**      **New genes for OI**

Moderator:    Antonella Forlino (Pavia, Italy)

**Invited speaker: Joan Marini (Bethesda, MD, US)**

What is Osteogenesis Imperfecta (OI) in 2017?

### Oral communications

**O32**

**Insights into bone phenotype and collagen biochemistry of TMEM38B null mutations causing Type XIV OI**

Nadja Fratzl-Zelman, Vienna Austria

**O34**

**Defective Regulated Intramembrane Proteolysis (RIP) due to mutations in MBTPS2 underlies X-linked Osteogenesis Imperfecta (OI), Joan Marini, Bethesda, MD, US**

**O33**

**Severe Osteogenesis Imperfecta presentation in a family with a novel CREB3L1 mutation**

Dimitra Micha, Amsterdam, The Netherlands

**O35**

**FAM46A mutations are responsible for autosomal recessive form of Osteogenesis Imperfecta**

Valerie Cormier-Daire, Paris, France

**10.45 - 11.10**

**Coffee break**

**11.10 - 11.30**

**Patient Organisations**

**Ingunn Westerheim (OIFE) & Tracy Hart (OIF)**

The role of the patient organisations

**11:30 – 12:30**

**Session 10**      **Consortia in OI**

Moderator:      Nick Bishop (Sheffield, UK)

**Invited speaker: Luca Sangiorgi (Bologna, Italy)**

European Reference Network for Rare Bone Disorders, ERN – BOND

**Invited speaker: Vernon R. Sutton (Texas, US)**

“The American way” - Brittle Bone Disorders Consortium

**Debate**

**12.45 - 13.00**

**Closing and final remarks**

*Announcement of the 14th International conference on OI*

**From 13.00**

**Grab & Go Lunch**