MECHANISTIC AND THERAPEUTIC ADVANCES IN RARE SKELETAL DISEASES

A meeting in affiliation with the American Society for Bone and Mineral Research (ASBMR)
This meeting is co-organized by the Rare Bone Disease Alliance, The Osteogenesis Imperfecta Foundation, and the NIH Brittle Bones Disorders Consortium

SEPTEMBER 26-27, 2018

Chair: Brendan Lee, MD, PhD, Baylor College of Medicine
Co-Chair: Maurizio Pacifici, PhD, Children's Hospital of Philadelphia

Program Committee:
Yang Chai, DDS, PhD, University of Southern California
Michael Collins, MD, National Institute of Dental and Craniofacial Research
Matthew Drake, MD, PhD, Mayo Clinic
Deborah Krakow, MD, UCLA and
Sandesh Nagamani, MD, Baylor College of Medicine

Meeting Agenda

Wednesday, September 26

7:00 am - 7:50 am **Breakfast**

7:50 am – 8:00 am **Welcome: Brendan Lee, MD, PhD, Baylor College of Medicine**

8:00 am - 9:40 am **Session 1: Diagnostic Approach to Rare Skeletal Diseases**
Moderator: Maurizio Pacifici, PhD, Children's Hospital of Philadelphia

Next Generation Sequencing and Multi-Omic Approaches for Diagnosing Skeletal Diseases
Brendan Lee, MD, PhD, Baylor College of Medicine

Higher Order Chromatin Structure and Distal Genetic Interactions in the Diagnosis of Skeletal Diseases
Struan Grant, PhD, Children's Hospital of Philadelphia

Radiographic and Ultrasound Imaging of Skeletal Diseases
Deborah Krakow, MD, University of California - Los Angeles

Past, Current and Future Biomarkers of the Skeleton
Charlotte Gistelinck, PhD, University of Washington

9:40 am - 9:55 am **Break**

10:00 am - 12:05 pm	**Session 2: Preclinical Models and Pathogenesis (Bones, Cartilage & Craniofacial)** Moderator: Deborah Krakow, MD, University of California - Los Angeles *Fibrous Dysplasia* Michael Collins, MD, National Institute of Dental and Craniofacial Research *BMP Signaling and Therapeutic Approaches* Aris Economides, PhD, Regeneron Pharmaceuticals and Dinko Gonzalez Trotter, PhD, Regeneron Pharmaceuticals *Osteogenesis Imperfecta* Frank Rauch, MD, Shriners Hospital of Montreal *Progressive Heterotopic Ossification* Yingzi Yang, PhD, Harvard School of Dental Medicine *Hereditary Multiple Exostoses* Maurizio Pacifici, PhD, Children's Hospital of Philadelphia
12:05 pm - 1:30 pm	**Lunch & Poster Session 1**
1:30 pm - 3:10 pm	**Session 2 Continued** Moderator: Yang Chai, DDS, PhD, University of Southern California *Fibrodysplasia Ossificans Progressiva* Eileen Shore, PhD, University of Pennsylvania *Craniosynostosis* Andrew Wilkie, FRS, FMedSci, FRCP, University of Oxford *Hypo-Oligodontia and Tooth Stem Cells* Ophir Klein, MD, PhD, University of California - San Francisco *Osteopetrosis* Anna Teti, PhD, University of L'Aquila
3:10 pm - 3:25 pm	**Break**
3:30 pm - 5:35 pm	**Session 3: Therapies on the Horizon and New Disease Targets** Moderator: Michael Collins, MD, National Institute of Dental and Craniofacial Research *Chemical Biology and Drug Discovery* Peter J. Brown, PhD, University of Toronto *Dental Stem Cell Therapies* Yang Chai, DDS, PhD, University of Southern California *Stem Cells for Treatment of OI and the BOOSTB4* Trial (sponsored by the ICCBH) Cecelia Gotherstrom, PhD, Karolinska Institutet

Gorham's Disease
Denise Adams, MD, Boston Children's Hospital

Melorheostosis
Timothy Bhattacharyya, MD, National Institute of Arthritis and Musculoskeletal and Skin Diseases

5:35 pm - 7:30 pm **Evening Reception/Poster Session 2**

Thursday, September 27

7:00 am - 8:00 am **Breakfast**

8:00 am - 9:40 am **Session 4: Targeting Signaling Pathways (Clinical)**
Moderator: Matthew Drake, MD, PhD, Mayo Clinic

The Clinical Trial of Anti-TGFb in OI
Sandesh Nagamani, MD, Baylor College of Medicine

The Role of Sclerostin Inhibition in Bone
Andreas Grauer, MD, Amgen

FGF23 and X-Linked Hypophosphatemia
Thomas Carpenter, MD, Yale University

C-Naturietic Peptide & Achondroplasia
Julie Hoover Fong, MD, PhD, Johns Hopkins University

9:40 am - 9:55 am **Break**

10:00 am - 12:05 pm **Session 5: Advances in Endpoints and Assessments (Preclinical & Clinical)**
Moderator: Sandesh Nagamani, MD, Baylor College of Medicine

HR-pQCT Evaluation of Bone
Steven Boyd, PhD, University of Calgary

MRI Evaluation of Cartilage
Sharmila Majumdar, PhD, University of California - San Francisco

FDA Approach to Novel Endpoints
Theresa Kehoe, MD and Gemma Kuijpers, PhD, FDA

Novel Growth Plate Markers
William Horton, MD, Oregon Health Sciences University

Hypophosphatasia: What's Next?
Michael Whyte, MD, Shriners Hospital - St. Louis

12:05 pm - 1:30 pm **Lunch and Poster Session 3**

1:30 pm - 2:45 pm	**Session 6: Current Industry Clinical Trials and Approach to Clinical Trials Phase 1 through Pivotal Phase 3 and Post-Approval Studies** Moderator: Michael Collins, MD, National Institute of Dental and Craniofacial Research *Burosumab Therapy in Children and Adults with XLH, from Discovery to Approval* Javier San Martin, MD, Ultragenyx *Addressing Scarcity and Heterogeneity in Drug Development for FOP* Scott Melis, MD, PhD, Regeneron *Fibrodysplasia Ossificans Progressiva: Use of a Natural History Study in the Design and Implementation of a Phase 3 Trial* Donna Grogan, MD, Clementia *Collaborative Use of a Patient Registry in the Design of a Phase 2 Study for a New Indication, Multiple Osteochondroma* Fei Shih, MD, PhD, Clementia *Clinical Development of Asfotase Alfa for the Treatment of Pediatric-Onset Hypophosphatasia* Tom Brown, PhD, Alexion
2:45 pm - 3:45 pm	**Industry and FDA Panel**: All speakers above and FDA Representatives Theresa Kehoe, MD and Gemma Kuijpers, PhD
3:45 pm	**Conclusion** – Brendan Lee, MD, PhD, Baylor College of Medicine