

Mechanistic and Therapeutic Advances in Rare Skeletal Diseases

A meeting jointly organized by the Rare Bone Disease Alliance (RBDA), a program of the Osteogenesis Imperfecta Foundation, and the Brittle Bone Disorders Consortium of the NIH Rare Bone Diseases Clinical Research Network to bring together scientists interested in rare bone disease. The program will cover state of the art topics in diagnosis, preclinical mechanisms of disease, clinical targeting of signaling pathways, and novel clinical endpoints for assessment of efficacy.

Program Chair: Brendan Lee, MD, PhD, *Baylor College of Medicine*

Program Co-Chair: Maurizio Pacifici, PhD, *Children's Hospital of Philadelphia*

Program Committee: Yang Chai, DDS, PhD, *USC*; Michael Collins, MD, *NIDCR*; Matthew Drake, MD, PhD, *Mayo Clinic*; Deborah Krakow, MD, *UCLA*; Sandesh Nagamani, MD, *Baylor College of Medicine*

The Rare Bone Disease Alliance recognizes the support of its program planning partner, International Conference on Children's Bone Health (ICCBH)

Preliminary Conference Agenda

Wednesday, September 26 – Morning

Session 1: Diagnostic Approach to Rare Skeletal Diseases

- Speaker 1:** Brendan Lee, MD, PhD (Baylor College of Medicine) on next generation sequencing and multi-omic approaches for diagnosing skeletal diseases.
- Speaker 2:** Struan Grant, PhD (Children's Hospital of Philadelphia) on higher order chromatin structure and distal genetic interactions in the diagnosis of skeletal diseases
- Speaker 3:** Deborah Krakow, MD (University of California, Los Angeles) on radiographic and ultrasound imaging of skeletal diseases
- Speaker 4:** David Eyre, PhD (University of Washington) on past, current and future biomarkers of the skeleton

Break

Session 2: Preclinical Models and Pathogenesis (Bones, Cartilage & Craniofacial)

Session 2A

- Speaker 1:** Michael Collins, MD (National Institute of Dental and Craniofacial Research) on Fibrous Dysplasia
- Speaker 2:** Frank Rauch, MD (Shriners Hospital of Montreal) on Osteogenesis Imperfecta
- Speaker 3:** Yingzi Yang, PhD (Harvard School of Dental Medicine) on Progressive Heterotopic Ossification
- Speaker 4:** Anna Teti, PhD (University of L'Aquila) on Osteopetrosis

Lunch & Poster Session 1

Session 2B – Afternoon

- Speaker 5:** Maurizio Pacifici, PhD (Children's Hospital of Philadelphia) on chondrodysplasias
- Speaker 6:** Andrew Wilkie, FRS, FMedSci, FRCP (University of Oxford) on craniosynostosis
- Speaker 7:** Ophir Klein, MD, PhD (University of San Francisco) on Hypo-oligodontia and tooth stem cells
- Speaker 8:** Peter J. Brown, PhD, (University of Toronto), on Chemical Biology and Drug Discovery

Light Dinner Fare & Poster Session 2 – Evening

Session 3: Therapies on the Horizon and New Disease Targets - Evening

- Speaker 1:** Yang Chai, DDS, PhD (University of Southern California) on dental stem cell therapies
- Speaker 2:** Cecelia Gotherstrom, PhD (Karolinska Institute) on Stem Cells for Treatment of OI and the BOOSTB4 Trial (sponsored by the ICCBH)
- Speaker 3:** Denise Adams, MD (Harvard) on Gorham's Disease
- Speaker 4:** Timothy Bhattacharyya, MD (National Institutes of Health) on Melorheostosis

Day 2 Thursday September 27 – Morning

Session 4: Targeting Signaling Pathways (Clinical)

- Speaker 1:** Sandesh Nagamani, MD (Baylor College of Medicine) on the clinical trial of anti-TGF β in OI
- Speaker 2:** Andreas Grauer, MD (Amgen) on anti-sclerostin therapy in OI
- Speaker 3:** Tom Carpenter, MD (Yale University) on FGF23 and X-linked hypophosphatemia
- Speaker 4:** Julie Hoover Fong, MD, PhD (Johns Hopkins) on C-natriuretic peptide & achondroplasia

Break

Session 5: Advances in Endpoints and Assessments (Preclinical & Clinical)

- Speaker 1:** Steven Boyd, PhD (University of Calgary) on HRPqCT evaluation of bone
- Speaker 2:** Sharmila Majumdar, PhD (University of San Francisco) on MRI evaluation of cartilage
- Speaker 3:** Theresa Kehoe, MD and Gemma Kuijpers, PhD (FDA) on FDA approach to novel endpoints.
- Speaker 4:** William Horton, MD (Oregon Health Sciences University) on novel growth plate markers

Lunch and Poster Session 3 – Afternoon

Session 6: Current Industry Clinical Trials and Approach to Pivotal Phase 3 and Post - Approval Studies

Presentations from Regeneron, Clementia, Ultragenyx, Mereo, Alexion, and other companies. Representatives will discuss status of ongoing clinical trials in rare bone diseases and post-marketing challenges. Panel discussion will focus on clinical trial design and endpoints.