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 (Karolina 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 Bhattachyrra, 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-TGFb 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-naturietic 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.