

## AGENDA

**Saturday, June 3rd**

**SESSION 1: Genetics: Where Are We Now?**

Moderator: Nick Bishop, MD

**Wolfgang Högler, MD (recorded presentation):** ALPL Gene Variant Database: Current Construction, Validation, and Interpretation

**Nilton Salles, MD, PhD:** Clinical, Biochemical, and Genetic Studies of Pediatric Hypophosphatasia “Carrier” Parents

**Michael P. Whyte, MD:** Implications of Positive Findings for Families

**SESSION 2: Mineralization, Genetics, and Epigenetics**

Moderator: Susan Ott, MD

**Carlos Ferreira, MD:** Pyrophosphate: Homeostasis and Aberrations

**Deborah Wenkert, MD:** The Phenome and HPP

**Marc McKee, PhD:** The Stenciling Principle for Mineralization: What is it and What is its Importance?

**SESSION 3: HPP in Adults**

Moderator: Tom Weber, MD

**Patrick Mantyh, PhD:** Pain: Is it in Your Muscles and/or Bones? What Explains Bone Pain?

**Gary Gottesman, MD:** Challenges of Diagnosing HPP: Molecular Diagnosis is Never Enough

**Peter Tebben, MD:** Hypophosphatasia or Osteoporosis: A potential clinical conundrum

**Chad Deal, MD:** Rheumatologic Manifestations of Hypophosphatasia (HPP)

**Lothar Seefried, MD:** HPP - What Have We Learned? Indications for Treatment?

## SESSION 4: HPP Beyond Bones & Joints

Moderator: Larry Suva, PhD

**Brian Foster, PhD:** Insights into Dentoalveolar Defects Associated with Hypophosphatasia from Novel Mouse Models

**Dana Gaddy, PhD:** Compromises in skeletal muscle and myoblast energetics are associated with compromised kinematics in sheep with hypophosphatasia (HPP)

**Victoria DeMambro, PhD Candidate:** Fat Metabolism in Hypophosphatasia: Clinical and Basic Implications

**Mark Rallo, OD:** Hypophosphatasia and Ectopic Calcification: Ocular Findings From the Enobia Study

**Eric Rush, MD:** Are there Phenocopies of HPP? (Yes!)

## SESSION 5: Treatment

Moderator: Alison Boyce, MD

**Richard Eastell, MD:** Biochemical Assessment of Adults in the Metabolic Bone Disease Clinic

**Laura Tosi, MD:** Orthopedics for Pediatric Hypophosphatasia

**Mark Nunes, MD:** Transition of Pediatric to Adult Care: Challenge and Opportunity

**Cheryl Rockman-Greenberg, MD (recorded presentation) and Kathryn Dahir, MD:** Enzyme Replacement Therapy Cessation in Adults with Hypophosphatasia

**Donna Griffin, DPT:** Monitoring of Physical Performance in Patients with Hypophosphatasia

**Tae Matsumoto, MD, PhD (Possibility of prenatal gene therapy for lethal HPP model mice)/  
Michael P. Whyte, MD:**  
Pregnancy? (Prenatal Treatment for Baby?)

**José Luis Millán, PhD (recorded presentation):**  
Gene Therapy for Hypophosphatasia

**Sunday, June 4th**

**SESSION 6: Research Roundup from Soft Bones Grant Recipients**

Moderator: Michael P. Whyte, MD

**Soft Bones Grant Recipients Update and Goals**

(individual 12 minute presentations)

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- Steve Mumm, PhD
  - Luke Mortensen PhD
  - Brian Foster, PhD
  - Kathryn Dahir, MD - (recorded presentation by Co-PI Michael de Riesthal, PhD, CCC-SLP)
  - Dobrawa Napierala, PhD
  - Dana Gaddy, PhD
  - Flavia Amadeu de Oliveira, PhD
  - Fatma F. Mohamed, PhD
  - Nan Hatch, DMD, PhD

**SESSION 7: Pharma Considerations for HPP Treatment**

Moderator: Suzanne Jan De Beur, MD

**Meeting Summary and Considerations for the Future**

Michael P. Whyte, MD

**Meeting Wrap Up and Conclusion  
(Attendee Comments and Suggestions)**

Michael P. Whyte, MD, Deborah Fowler