

## PROGRAM

*The scientific sessions will be held in the Suffolk Room.*

### THURSDAY, OCTOBER 29

19:00-22:00            **Welcome reception (Suffolk Room)**

### FRIDAY, OCTOBER 30

7:30-8:30            *Continental Breakfast (Norfolk Room)*

8:30-8:40            **Opening Remarks**

#### **Session 1: Overview and Clinical Presentation of MHE** (Discussion Leader: Dror Paley)

8:40-9:00            **Dan Wells** (University of Houston)  
*Perspectives on the Genetics of Multiple Hereditary Exostosis*

9:00-9:30            **Dror Paley** (Paley Advanced Limb Lengthening Institute)  
*Multiple Osteochondromas Treatment of the Lower Limb Deformities*

9:30-10:00          **Scott Kozin** (Shriners Hospital for Children, Philadelphia)  
*MHE of the Upper Extremity*

10:00-10:20        *Coffee Break*

#### **Session 2: Human Genetics of MHE** (Discussion Leaders: Wim Wuyts and Luca Sangiorgi)

10:20-10:50        **Wim Wuyts** (University of Antwerp)  
*Genetics of Multiple Osteochondromas: Overview of the Current Status*

10:50-11:10        **Ivy Jennes** (University of Antwerp)  
*Characterisation of the Promoter Region of the Human Exostosin-1 Gene*

11:10-11:40        **Luca Sangiorgi** (Rizzoli Orthopaedic Institute)  
*Genotype-Phenotype Correlation Study in 529 MO Patients: 'Protective' and 'Risk' Factors*

11:40-12:00        **Elena Pedrini** (Rizzoli Orthopaedic Institute)  
*Osteochondroma Onset and Malignant Degeneration: Redefinition of EXT Gene Role*

12:00-12:10        **Judith Bovée** (Leiden University)  
*Osteochondroma Formation: Haploinsufficiency or Two Hits*

12:10-13:30        *Lunch (Norfolk Room)*

**Session 3: Biochemistry, Chemistry, and Cell Biology of Heparan Sulfate** (Discussion Leader: Marion Kusche-Gullberg)

- 13:30-14:00      **Marion Kusche-Gullberg** (University of Bergen)  
*EXT-Dependent Regulation of Heparan Sulfate Structure and Function*
- 14:00-14:30      **Robert Linhardt** (Rensselaer Polytechnic Institute)  
*New Advances in Heparan Sulfate/Heparin Analysis and Biosynthesis*
- 14:30-15:00      **Jeremy Turnbull** (University of Liverpool)  
*Elucidating the Functions of Heparan Sulfates: Towards Glycomics Strategies*
- 15:00-15:20      **Cathy Merry** (University of Manchester)  
*Restoring Function to Heparan-Sulphate Deficient Cells*
- 15:20-15:40      *Coffee Break*

**Session 4: Studies Using Non-Mammalian Model Animals** (Discussion Leader: Henry Roehl)

- 15:40-16:10      **Henry Roehl** (University of Sheffield)  
*Zebrafish as a Model for Multiple Hereditary Exostoses*
- 16:10-16:40      **Rahul Warrior** (University of California, Irvine)  
*Developmental Regulation of Heparan Sulfate Proteoglycan Synthesis*
- 16:40-17:10      **Joseph Yost** (University of Utah)  
*HS Fine Structure and FGF Signaling Pathways Converge at Cilia: Does Cilia Function Have a Role in HME?*
- 17:10-17:30      **Malgorzata Wiweger** (Leiden University)  
*New Aspects of Multiple Osteochondromas – A Lesson from dackel (dak/ext2) Zebrafish Mutant*

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19:30-22:30      *Dinner (Suffolk Room)*

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## **SATURDAY, OCTOBER 31**

8:00-9:00 *Continental Breakfast (Norfolk Room)*

### **Session 5: Developmental Biology of Bone and Cartilage** (Discussion Leader: Henry Kronenberg)

- 9:00-9:30 **Henry Kronenberg** (Massachusetts General Hospital)  
*PTH/PTHrP Receptor Signaling is Required for Maintenance of the Growth Plate in Postnatal Life*
- 9:30-10:00 **David Ornitz** (Washington University School of Medicine)  
*FGF Signaling in Skeletal Development and Repair*
- 10:00-10:20 **Patrick Allard** (Harvard Medical School)  
*Understanding the Role of Ihh and Proteoglycans Interaction during Bone Development*
- 10:20-10:40 *Coffee Break*
- 10:40-11:10 **Michael Underhill** (University of British Columbia)  
*Regulation of Osteogenesis by the Retinoic Acid Signaling Pathway*
- 11:10-11:40 **Olena Jacenko** (University of Pennsylvania)  
*Altered Matrix at the Chondro-Osseous Junction Leads to Defects in the Hematopoietic Stem Cell Niche*
- 11:40-12:00 **Kyle Kurek** (Children's Hospital, Boston)  
*Metachondromatosis: Expanding the Clinicopathologic Spectrum*
- 11:50-1:30 *Lunch (Norfolk Room)*

### **Session 6: Role of Heparan Sulfate in Bone Development / Novel MHE Mouse Models** (Discussion Leader: Maurizio Pacifici)

- 13:30-14:00 **Maurizio Pacifici** (Thomas Jefferson University)  
*Mechanisms of Exostosis Formation in Mouse Models of HME*
- 14:00-14:30 **Kevin Jones** (University of Utah)  
*Osteochondromagenesis: Somatic Loss of Heterozygosity Modeled via Cre-Mediated Inversion of the Second Exon of Ext1 in Chondrocytes*
- 14:30-15:00 **Andrea Vortkamp** (University of Duisburg-Essen)  
*Molecular Characterization of Osteochondroma Development in Mice*
- 15:00-15:20 **Kazu Matsumoto** (Burnham Institute for Medical Research)  
*Stochastic Conditional Knockout of Ext1 Reveals an Unexpected Relationship between Biallelic Inactivation of the Gene and the*

*Development of Multiple Exostoses*

15:20-15:40      *Coffee Break*

**Session 7: Role of Heparan Sulfate in Non-Skeletal Tissues** (Discussion Leader: Jeffrey Esko)

15:40-16:10      **Jeffrey Esko** (University of California, San Diego)  
*Do Mutations in EXT1 or EXT2 Affect Non-Skeletal Tissues?*

16:10-16:40      **Hudson Freeze** (Burnham Institute for Medical Research)  
*Deficient Heparan Sulfate and N-Glycosylation Contribute to Protein-Losing Enteropathy in Humans and Mice*

16:40-17:10      **Yu Yamaguchi** (Burnham Institute for Medical Research)  
*Deficiency of Heparan Sulfate in Excitatory Neurons Causes Autism-like Behaviors in Mice*

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19:30-22:30      *Dinner Banquet (Suffolk Room)*

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**SUNDAY, NOVEMBER 1**

8:00-8:30      *Continental Breakfast (Suffolk Room)*

**Session 8: Biology of MHE and Related Bone Disorders** (Discussion Leader: Benjamin Alman)

8:30-9:00      **Benjamin Alman** (University of Toronto)  
*Gli2 and p53 Cooperate to Regulate IGFBP-3–Mediated Chondrocyte Apoptosis in the Progression from Benign to Malignant Cartilage Tumors*

9:00-9:30      **Pancras Hogendoorn** (Leiden University)  
*Primary Cilia Organization Orchestrating Cell Polarity in the Growth Plate and its Loss in Osteochondroma*

9:30-10:00      **Frederick Kaplan** (University of Pennsylvania)  
*Osteochondromas & the FOP Metamorphogene*

10:00-10:30      **Judith Bovée** (Leiden University)  
*On the Clinical Manifestation and the Genetics of Ollier Disease*

10:30-11:15      **Meeting Summary and General Discussion** – Discussion Leaders: Yu Yamaguchi and Maurizio Pacifici  
**Remarks from the MHE Research Foundation** – Sarah Ziegler

11:15      *Departure*