

## Skeletal Dysplasia Group Spring Meeting 2019

### New advances in the treatment of skeletal dysplasia: trials and tribulations

**Date:** Friday 5<sup>th</sup> April

**Venue:** Prospero House, 241 Borough High Street, London SE1 1GA

**Programme:**

**09.30-10.00 Registration & Refreshments**

**10.00-10.10 Welcome**

Melita Irving

Consultant in Clinical Genetics

Guy's and St Thomas' Hospital, London

**10.10-10.40 Overview of 'druggable' targets in skeletal dysplasia**

Mike Briggs

Professor of Skeletal Genetics

University of Newcastle

**10.40-11.40 Achondroplasia:**

**10.40-11.10 C-Natriuretic peptide: the long and short of it**

Melita Irving

Consultant in Clinical Genetics

Guy's and St Thomas' Hospital, London

**11.10-11.40 Other FGFR3 targets: Decoys and destroyers**

Christine Burren

Consultant in Paediatric Diabetes, Endocrinology and Bone

Bristol Royal Hospital for Children

**11.40-12.00 Grant application updates:**

**Whole exome sequencing in skeletal dysplasia**

Melita Irving/Ana Belez

Consultant in Clinical Genetics

Guy's and St Thomas' Hospital, London

**Management guidelines in adults with achondroplasia**

Michael Wright/Marta Bartoli

Consultant in Clinical Genetics, Newcastle Upon Tyne Hospitals

- 12.00-12.30**    **Unknown cases**
- 12.30-13.20**    **Lunch**
- 13.20-13.40**    **Multiple osteochondromas: MO-PED Vavavoom!**  
Mars Skae  
Consultant in Paediatric Endocrinology  
Royal Manchester Children's Hospital
- 13.40-14.00**    **X-Linked hypophosphatasia: Burosumab comes to town!**  
Moirra Cheung  
Consultant in Paediatric Endocrinology and Rare Bone Disease
- 14.00-14.20**    **The asfotase alpha to omega of hypophosphatasia**  
Vrinda Saffaf  
Consultant in Paediatric Endocrinology  
Birmingham Women's and Children's Hospital
- 14.40-15.00**    **Schmid metaphyseal dysplasia: a tale in drug repurposing**  
Marta Bartoli/Michael Wright  
Consultant in Clinical Genetics  
Newcastle Upon Tyne Hospitals
- 15.00-15.20**    **Tea & Refreshments**
- 15.20-15.50**    **Findacure: bringing the rare disease community together**  
TBC
- 15.50-16.30**    **Horizon scanning – panel discussion**  
Convener: Melita Irving
- 16.30**            **Close**

*(CPD points: applied for)*