

Skeletal Dysplasia Group Spring Meeting 2019

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

Date: Friday 5th 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 Beleza

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: Barosumab comes to town!

Moira Cheung

Consultant in Paediatric Endocrinology and Rare Bone Disease

14.00-14.20 The asfotase alpha to omega of hypophosphatasia

Raja Padidela

Consultant in Paediatric Endocrinology

Manchester University Foundation Trust

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

Rick Thompson

15.50-16.10 Hajdu-Cheney Syndrome support group – the power of the patient to drive research

HJCS support group rep

16.10-16.30 Horizon scanning – panel discussion

Convener: Melita Irving

16.30 Close

(CPD points: applied for)

