

SDG Autumn Meeting 2022
MC and MI Dots and Dashes
November 10th (pm) and 11th November (am)

Thursday November 10th pm Session - Dashes (calcification)

13.00pm	Introduction
13.10pm	Hyperphosphatemic tumoral calcinosis <i>Dr Rachel Gafni -Senior Research Physician, Skeletal Disorders and Mineral Homeostasis Section, National Institute of Dental and Craniofacial Research National Institutes of Health, USA</i>
13.40pm	Radiology of extraskeletal calcification <i>Dr Ali Calder - Consultant Radiologist, Great Ormond Street Hospital, UK</i>
14.10 pm	Break
14.15pm	ENPP1/GACI <i>Dr Carlos Ferreira - Head, Skeletal Genomics Unit, National Human Genome Research Institute, National Institutes of Health, USA</i>
14.45pm	Fibrodysplasia ossificans progressiva <i>Dr Genevieve Baujat - Clinical Geneticist Consultant, Imagine Institute of Necker-Enfants Malades Hospital, Paris, France</i>
15.15pm	Break
15.20pm	GNAS ectopic ossification <i>Dr Anya Rothenbuhler - Pediatric Diabetes and Endocrinology department Reference Center for rare Diseases of Calcium and Phosphate Metabolism Kremlin Bicêtre University Hospital</i>
15.50pm	Reports back of travel grants (5 mins per presentation)
16.30pm	AGM



Friday November 11th am Session Dots (stippling)

09.00am	Welcome
09.05am	Clinical and Genetic aspects of stippling conditions <i>Dr Ataf Sabir - Consultant in Clinical Genetics, Birmingham Women and Children's Hospital</i>
09.45am	Radiology in stippling condition <i>Dr Rike Schiller - Fellow, Paediatric Radiology INSELSPITAL, Universitätsspital Bern</i>
10.15am	Break
10.30am	Stippling and the cholesterol pathway <i>Dr Max Rossi - Consultant in Clinical Genetics, Hospices Civils de Lyon, France</i>
11:00am	Gene hunting in unexplained skeletal dysplasia with chondrodysplasia punctata <i>Dr Alistair Pagnamenta - post-doctoral researcher, Wellcome Centre for Human Genetics, Oxford</i>
11:30am	Break
11.45am	Unknown Cases and 100K solved cases <i>Moderator: Dr Melita Irving - Consultant in Clinical Genetics, Guy's and St Thomas' NHS Trust, London</i>
12:30pm	Rhizomelic chondrodysplasia punctata <i>Dr Mike Bober - Consultant in Clinical Genetics, Wilmington, Delaware</i>
13.00pm	Close of meeting



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