



TRANSLATIONAL RESEARCH IN MONOGENIC BONE DISORDERS: FROM BENCH TO BEDSIDE

SESSION 1	
RARE MONOGENIC BONE DISORDERS – WHAT’S NEW (Moderator: Jannie Dahl Hald, Aarhus University, DK)	
09.00 – 09.10	OPENING Outi Mäkitie, University of Helsinki, FI
09.10 – 09.40	ACHONDROPLASIA: LATEST UPDATES WITH A FOCUS ON DRUG DISCOVERY Melita Irving, NHS Trust, UK
09.40 – 10.10	ANIMAL MODELS OF OSTEOPETROSIS Antonio Maurizi, University of L'Aquila, IT
10.10 – 10.40	X-LINKED HYPOPHOSPHATEMIA: FROM DISEASE PATHOGENESIS TO IMPROVED PATIENT CARE Justine Bacchetta, Femme Mère Enfant Hospital, FR
10.40 – 11.00	DISCUSSION
11.00 – 11.15	BREAK
SESSION 2	
FROM RARE BONE DISORDERS TO POPULATION STUDIES (Moderator: Evangelia Ntzani, University of Ioannina, GR)	
11.15 – 11.45	OI SPECTRUM: FROM GENETIC DISCOVERIES TO NOVEL PATHWAYS Emma Duncan, King’s College, UK
11.45– 12.15	NEW THERAPIES OF GENETIC BONE DISORDERS: CRISPR/CAS9 GENE EDITING AND CELL THERAPY IN THE TREATMENT OF OSTEOGENESIS IMPERFECTA - WHERE ARE WE NOW Fleur van Dijk, NHS Trust, UK
12.15–12.35	FROM FAMILIES TO POPULATION STUDIES: PUTTING TOGETHER A STUDY COLLECTION Melissa M Formosa, University of Malta, MT
12.35 – 12.45	DISCUSSION
12.45 – 13.30	BREAK
SESSION 3	
CASE PRESENTATIONS OF MONOGENIC BONE DISORDERS (Moderator: Maria Carolina Medina Gomez, Erasmus MC, NL)	
13.30 – 13.45	RARE VARIANT ANALYSIS IN BONE METABOLISM GENES IN A TRUNCATED COHORT OF POST-MENOPAUSAL WOMEN Kelly Rabionet, University of Barcelona, ES
13.45 – 14.00	A HOMOZYGOUS MISSENSE VARIANT IN SP7/OSTERIX CAUSES A NEW FORM OF CRANIODIAPHYSEAL DYSPLASIA Gretl Hendrickx, KU Leuven, BE
14.00 – 14.15	WNT PATHWAY VARIANTS AT THE FRONTIER BETWEEN MONOGENIC AND POLYGENIC BONE DISORDERS José Riancho, University of Cantabria, ES
14.15 – 14.30	KIF24 VARIANTS AS A NOVEL CAUSE OF SKELETAL DYSPLASIA Outi Mäkitie, University of Helsinki, FI
14.30 – 14.50	DISCUSSION
14.50 – 15.10	BREAK
SESSION 4	
CELLS, ANIMAL MODELS & BEYOND – USING DIFFERENT TOOLS TO IDENTIFY NEW GENES & DRUG TARGETS IN BONE DISORDERS (Moderator: Dylan JM Bergen, University of Bristol, UK)	
15.10 – 15.30	TISSUE ENGINEERING SYSTEMS AS TRANSLATIONAL MODELS IN SKELETAL DISORDERS Ivan Duran, University of Malaga, IB
15.30 – 15.50	THE MUSCULOSKELETAL KNOWLEDGE PORTAL Douglas Kiel, Harvard Medical School, US
15.50 – 16.10	USING ZEBRAFISH TO VALIDATE OSTEOANABOLIC TARGETS Georgina McDonald, University of Bristol, UK
16.10 – 16.25	DISCUSSION
16.30	CLOSING

