Preliminary Programme

Thursday 21 September 2017

09h00 – 10h30: Session 1: New phenotypes and Genes

ISDS/038   Mutations in C-Natriuretic Peptide (CNP): a novel cause of autosomal dominant short stature and brachydactyly. Heath KE

ISDS/083   Mutations in fibronectin cause a subtype of spondylometaphyseal dysplasia with “corner fractures” Lee CS

ISDS/032   Gain-of-function mutation in a novel gene causes a novel human spondyloepimetafhyseal dysplasia Grigelioniene G.

ISDS/026   Axial spondylometaphyseal dysplasia- a specific form of skeletal ciliopathy with growing complexity Ikegawa S

ISDS/156   NANS-mediated synthesis of sialic acid is required for brain and skeletal development Bonafe L.

ISDS/123   Further delineation of spondyloepimetafhyseal dysplasia faden-alkuraya type: a rspry1-associated spondylo-epi-metaphyseal dysplasia with cono-brachydactyly and craniosynostosis Simsek-Kiper

11h00 – 12h30: Session 2: Lessons from animal models

ISDS/077   proteoglycan synthesis defects in an in vivo model of desbuquois dysplasia type 1 Paganini C

ISDS/149   Investigating the SEMDJL disease causing mutations in Kif22 in skeletal development and disease Pirog K.A

ISDS/113   The role of Creld2 in skeletal development and disease. Dennis E. P

ISDS/091   Using patient derived induced pluripotent stem cells to model multiple epiphyseal Dysplasia Steven Woods1

ISDS/043   The disease mechanisms of skeletal dysplasia caused by two aggrecan mutations Gibson B.G

ISDS/041   Does a matrilin-3 mutation (p.T298M) knock-in mouse model mimic human osteoarthritis? Zaucke F

13h30 – 15h15: Session 3: NGS in large cohorts

ISDS/033   Penelope and the skeleton: Value of an undiagnosed disease program in the diagnosis, discovery and care of children with genetic bone disorders - Botto L.D

ISDS/109   Comprehensive clinical and genomic analysis of a large skeletal dysplasia cohort Alhashem Amal
**ISDS/111**  
*Use and efficiency of targeted NGS panel in skeletal dysplasias: experience on 330 patients* - Michot C.

**ISDS/117**  
*High succes in molecular studies among 215 Skeletal Dysplasias in Brazil*  
Cavalcanti D.P

**ISDS/069**  
*Next-generation diagnostic service for skeletal dysplasia diagnosis - our experience* - Beleza-Meireles A

**ISDS/134**  
*Results of the analysis of 370 probands using a Skeletal Dysplasia Next-Generation Sequencing panel* - Barraza-Garcia J.

**ISDS/052**  
*A decade of experience of molecular testing for skeletal dysplasia in India*  
Girisha Katta

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**Friday 22 September 2017**

**08h30 – 09h45: Session 4: Cellular and animal models**

**ISDS/062**  
*Cellular response to mutant collagen type I in patients with osteogenesis imperfecta can be a novel therapeutic target* - Besio R.

**ISDS/031**  
*Misregulation of a chaperone complex that modulates lysyl hydroxylation of Type I procollagen causes Osteogenesis Imperfecta* - Duran, I.

**ISDS/157**  
*Mutations in LRP4 can cause sclerosteosis in human and in mice*  
Boudin E

**ISDS/102**  
*Activating FGFR3 mutation in osteoblast affects appendicular and cranio-facial skeleton development* - Biosse Duplan M

**ISDS/061**  
*Longitudinal bone growth velocity assessment by near-infrared imaging in a murine model of achondroplasia* - Florence Authier

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**10h15 – 11h45: Session 5: Genes and phenotypes**

**David Rimoin lecture:**  
*Cartilage – selective Gene Expression and mechanisms of disease in the Skeletal Dysplasias* -- Daniel Cohn

**ISDS/112**  
*Clinical and Radiological characterization of EXTL3-related Skeletal Phenotype*  
Sousa S.B

**ISDS/154**  
*Exostosin-like 3 (EXTL3) deficiency: an autosomal recessive condition that impairs synthesis of heparan sulfate and affects bone, brain and the immune system*  
S. Volpi

**ISDS/118**  
*New genes for Robinow syndrome allow genotype-phenotype correlations that inform prognosis and gene function* - Sutton, V.R.

**ISDS/063**  
*Brachyolmia resulting from mutations in PAPSS2*  
Smithson S
ISDS/021  Systematic Phenotypic Characterisation of Skeletal Dysplasias with the Human Phenotype Ontology - Zankl Andreas

16h00 – 17h00: Session 6: Severe perinatal disorders

ISDS/092  Jeune thoracic dysplasia/short rib-polydactyly type III: clinical and molecular review of 125 cases - Cormier-Daire V

ISDS/098  previously unrecognized lethal dysostoses
Nishimura Gen

ISDS/132  A new proposed classification of Perinatal Lethal Hypophosphatasia after introduction of enzyme replacement therapy - Muotsuki J

ISDS/010  Novel Imaging Techniques in Skeletal Dysplasias: The use of Micro-CT
Shelmerdine S

17h30 – 19h00: Session 7: More on phenotype and natural history

Filip Vanhoenacker

ISDS/024  Mucolipidosis III Gamma: Clinical characterization and molecular analysis in 17 patients from India, Turkey and North America - Nampoothiri S

ISDS/034  Intermediate Autosomal Recessive Osteopetrosis: Long-term Follow up on 3 cases with CLCN7 mutations - Carminho-Rodrigues T

ISDS/099  Achondroplasia Natural History: the power of a multi-center clinical study
Hoover-Fong JE

ISDS/040  Disruptive, targeted emerging therapies in skeletal dysplasias.
Savarirayan, R

ISDS/153  Deficiency of sFRP4, a soluble LRP receptor antagonist, impairs the formation of cortical bone and results in Pyle disease - Pelin Ozlem Kiper-Simsek

Saturday 23 September 2017

09h00 – 10h30: Session 8: Animal models for treatment

ISDS/059  Novel therapeutic interventions for pseudoachondroplasie
Hecht

ISDS/146  Stimulating intracellular proteolysis reduces disease severity in an ER stress-related chondrodysplasia - Boot-Handford, Ray

ISDS/070  Use of chemical chaperones to target cellular stress in chihuahua, a zebrafish model of dominant osteogenesis imperfecta - Tonelli F

ISDS/054  Efficacy of palovarotene oral treatment on prevention of osteochondroma formation in the Fsp1-Ext1 conditional knockout mouse model of multiple osteochondromas - Lemire I
Oral administration of meclozine for the treatment of short stature in achondroplasia - Matsushita M

FLAG-sFGFR3 treatment prevents the metabolic deregulations in achondroplasia
Celine Saint-Laurent

11h00 – 12h15: Session 9: Treatment: ready for patients?

Invited lecture: Tissue Engineering for the Healing of Large Bone Defects.
Frank Luyten

Biomarin Corporate Symposium

Hajdu-Cheney syndrome: current treatments and drug repositioning strategies in severe osteoporosis - Irving M

Results from a Randomized, Placebo-Controlled, Double-Blind Study of Palovarotene in Subjects with Fibrodysplasia Ossificans Progressiva (FOP)
Kaplan F