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 spondyloepimetaphyseal 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 spondyloepimetaphyseal dysplasia faden-alkuraya type: a rspry1-associated spondylo-epi-metaphyseal dysplasia wıth 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

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

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 -** Murotsuki 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 Autossomal 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

ISDS/152 **Oral administration of meclozine for the treatment of short stature in achondroplasia** - Matsushita M

ISDS/030 **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 Coporate Symposium

ISDS/119 **Hajdu-Cheney syndrome: current treatments and drug repositioning strategies in severe osteoporosis** - Irving M

ISDS/047 **Results from a Randomized, Placebo-Controlled, Double-Blind Study of Palovarotene in Subjects with Fibrodysplasia Ossificans Progressiva (FOP)** Kaplan F