

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 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

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 **Mucopolidosis 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 pseudoachondroplasia**
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 Corporate 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