PRELIMINARY PROGRAM V.3
ISDS MEETING 2022

Local time GMT-4	**Wednesday 24th August, 2022**
16:00 - 18:00	**Registration (Hotel Best Western Premier Marina Las Condes)**
18:30 - 21:00	**Welcome reception**

Local time GMT-4	**Thursday 25th August, 2022**
07:00 - 08:30	**Registration (Hotel Best Western Premier Marina Las Condes)**
08:30 - 08:45	**Welcome - Dr. Ignacio Sánchez - President of Pontificia Universidad Católica de Chile**
08:45 - 10:30	**Session 1: Molecular mechanisms and phenotypes 1** | Moderators: Carlos Bacino - Andreas Zankl
08:45	**ISDS-115, Lianlei Wang** Disruptions of *SOX9* K2 domain reduce protein stability and induce mild skeletal dysplasia and late-onset scoliosis
09:00	**ISDS-17, Valerie Cormier-Daire** Biallelic variants in *SLC35B2* cause a novel chondrodysplasia with hypomyelinating leukodystrophy
09:15	**ISDS-19, Guilherme Yamamoto** Rothmund-Thomson syndrome type I with growth hormone deficiency is associated with a not previously described gene in an autosomal recessive inheritance pattern in seven families
09:30	**ISDS-114, Annette Madison** Clinical and molecular characterization of Chilean patients with X-linked hypophosphatemia
09:45	**ISDS-61, Gretl Hendrickx** Heterozygous pathogenic variants in the *CBFB* gene cause a novel phenotype resembling cleidocranial dysplasia
10:00 - 10:30	**ISDS-126, Andrea Superti-Furga** MutLands, a new advancement in the anatomic pathology of the human genome and in variant interpretation
10:30 - 11:00	Coffee break
11:00 - 12:15	**Session 2: Molecular mechanisms and phenotypes 2** | Moderators: Valérie Cormier-Daire - Antonio Rossi
11:00	**ISDS-02, V. Reid Sutton** Quantitative phenotypic analyses of Robinow syndrome: WNT signalling perturbation and phenotypic similarities and variability
11:15	**ISDS-16 Nathalia Liberatoscioli Menezes de Andrade** High prevalence of growth plate related genes in a cohort of children with isolated short stature
11:30	**ISDS-25 Sarah Smithson** Rare genetic mechanisms in skeletal dysplasia solved by reanalysis of whole exome/genome data
11:45	**ISDS-82 Lorenzo Botto** More than meets the eye: unsuspected systemic diseases identified through skeletal dysplasias.
12:00	**ISDS-111, Andreas Zankl** Integrating the ISDS Nosology with the G2P Project and the MONDO Disease Ontology
12:15 - 12:45	**David Rimoin lecture, Professor Matthew Warman**
13:00 - 14:00	Lunch
14:00 - 14:20	**Biomarin Corporate Symposium**
14:20 - 15:20	**Session 3: focus on molecular diagnosis 1** | Moderators: Karen Heath - Geert Mortier
14:20	**ISDS-68, Daniel Carvalho** TAR syndrome cohort of 20 Brazilian patients: variable clinical phenotype and molecular characterization of *RBM8A* variants.
14:35	**ISDS-76, Yiming Wang** Development of a Genetic Diagnostic Algorithm for Individuals with Split Hand Foot Malformation
14:50	**ISDS-130, Carlos Bacino** Increasing the Diagnosis Rate in Rare Bone disorders. Our Experience in the Undiagnosed Disease Network at Baylor College of Medicine
15:05	**ISDS-14, Alistair Calder** Eiken dysplasia with Parathyroid Hormone resistance: two new paediatric cases with the same novel variant
15:20	**ISDS-41, Bruno Marcarini** Next-generation sequencing applied to skeletal disorders: a ten-year experience in a Tertiary Center in Brazil
15:35 - 15:50	Coffee break
15:50 - 16:50	**Poster session 1**
17:10	**Social program - free evening**

Local time GMT-4	**Friday 26th August, 2022**
08:30 - 10:00	**Session 4: clinical series and observations** | Moderators: Alistair Calder - Ravi Savarirayan
08:30	**ISDS-09, Sheela Nampoothiri** Skeletal Dysplasia Registry from a Tertiary Hospital from South India : 16 Years Experience
08:45	**ISDS-35, Isabela Pasa** Molecular analysis of severe cases of Osteogenesis imperfecta and other rare low-mineral density disorders
09:00	**ISDS-44, Maria Eduarda Gomes** Ancestry study in Brazilian patients with Cartilage Hair Hypoplasia syndrome with possible founder effect of the g.196C>T variant
09:15	**ISDS-53, Cathleen Raggio** (To be confirmed) Should osteogenesis imperfecta be labeled as a low bone mass condition?
09:30	**ISDS-13, Caroline Michot** Nine new cases of spondylometaphyseal dysplasia with corner fractures: enhancement of the phenotypic spectrum of *FN1* gene mutations
09:45	**ISDS-52, Lucy Scrimshaw** Cerebral vasculopathy in osteopathia striata with cranial sclerosis, a rare but important complication.
10:00 - 10:45	Coffee break
10:45 - 11:30	**Session 5: skeletal dysplasias: prenatal aspects** | Moderators: Melita Irving - Denise Cavalcanti
10:45	**ISDS-43, Rhoda Akilapa** Observational study of fetal foramen magnum, thoracolumbar kyphosis and hippocampal development in Achondroplasia
11:00	**ISDS-62, Catherine Gooch** Diagnostic challenges of fetal skeletal dysplasia: A cohort of patients with prenatal diagnoses of skeletal dysplasias with unique or atypical postnatal courses
11:15	**ISDS-78, Nicole Nakousi-Capurro** A cohort of Brazilian OI patients with predominance of lethal phenotypes and analysis of the known recurrent mutations in Collagen 1 genes
11:30 - 12:00	**Lecture on Nosology**
12:00 - 13:00	**ISDS Business Meeting**
13:00 - 14.00	Lunch
13:10 - 13:30	**Takeda Corporate Lunch Symposium**
14.00 - 15.15	**Session 6: Evaluation of growth, quality of life, pain and functionality** | Moderators: Roberto Mendoza - Jacqueline Hecht
14:00	**ISDS-54, Antonio Rossi** Identification of potential non-invasive biomarkers in diastrophic dysplasia
14:15	**ISDS-66, Ximena Ortega** Diffusion-Tensor Imaging of the Physes: new technique to evaluate skeletal growth in patients with skeletal dysplasia
14:30	**ISDS-42, Penelope Ireland** Development of the Screening Tool for Everyday Mobility and Symptoms (STEMS) for Skeletal Dysplasia
14:45	**ISDS-70, Rosario Ramos Mejia** Evaluation of functionality-mobility in patients with skeletal dysplasias in Argentina: application of the STEMS (Screening Tool for Everyday Mobility and Symptoms)
15:00	**ISDS-105, Natassja Billich** Nutritional issues in skeletal dysplasia: A scoping review
15:15 - 15:45	Coffee break
15:45 - 17:00	**Session 7: Biomarkers and mouse models** | Moderators: Matthew Warman - Caroline Michot
15:45	**ISDS-75, Jacqueline Hecht** Nutraceutical treatment strategies in a mouse model of pseudoachondroplasia
16:00	**ISDS-48, Elvire Gouze** Lentiviral vector-based gene therapy for type II collagen disorders
16:15	**ISDS-64, Pavel Krejci** Modelling achondroplasia in mouse to address the future therapy targets
16:30	**ISDS-95, Laurence Legeai-Mallet** Hypochondroplasia gain-of-function mutation in *FGFR3* causes defective bone mineralization in mice
16:45	**ISDS-20,Carl Dambkowski** Low-dose infigratinib, an oral selective fibroblast growth factor receptor tyrosine kinase inhibitor, demonstrates activity in a preclinical model of hypochondroplasia
17:00 - 18:00	**Poster session 2**
19:15 - 23:00	**Conference dinner**
21:00	**Maroteaux Award**

Local time GMT-4	**Saturday 27th August, 2022**
08:30 - 09:30	**Patient family session**
09:45 - 11:15	**Session 8: Clinical and observational studies** | Moderators: Debora Bertola - Lorenzo Botto
09:45	**ISDS-107, Ilian Arsof** Quantifying Outcomes in Fibrodysplasia Ossificans Progressiva (FOP) by Patient Age: Results from an International Burden of Illness Survey
10:00	**ISDS-39, Melita Irving** ACHieve Study: Baseline Characteristics of a Multicenter Observational Study of Children with Achondroplasia
10:15	**ISDS-10, David Tunkel** Otolaryngology Utilization in Patients with Achondroplasia: Results from the CLARITY Study
10:30	**ISDS-28, Juan Llerena Jr** Health-related quality of life (HRQoL) in achondroplasia: findings from LISA (Life Impact Study on Achondroplasia), a multinational and observational study in Latin America
10:45	**ISDS-33, Ravi Savarirayan** Medical history of children enrolled in PROPEL: A prospective clinical assessment study in children with achondroplasia
11:00	**ISDS-119, Julie Hoover-Fong** Mode of Delivery Influence on Surgical Morbidity in Fetuses with Achondroplasia from CLARITY (Achondroplasia Natural History Study)
11:15 - 11:35	**Ipsen Corporate Symposium** "FOP Clinical and molecular diagnosis, and genotype-phenotype correlation"
11:35 - 12:00	Coffee break
12:00 - 12:45	**Session 9: Treatment Trials for Achondroplasia** | Moderators: Julie Hoover-Fong - Reid Sutton
12:00	**ISDS-45, Nadia Merchant** A Prospective Clinical Trial of Vosoritide in Hypochondroplasia: Baseline Demographics and Preliminary Results
12:15	**ISDS-69, Ravi Savarirayan** A Randomized Controlled Trial of Vosoritide in Infants and Toddlers With Achondroplasia
12:30	**ISDS-97, Melita Irving** Insights into the effects of vosoritide on the craniofacial structure of children with achondroplasia
12:45 - 13:00	ISDS Awards Ceremony - Organizing committee Closing remarks
13:00 - 14:00	Lunch