ISDS

PROGRAM

ISDS MEETING 2022

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

Local time GMT-4	**Thursday 25th August, 2022**
08:00 - 08:30	**Registration (Best Western Premier Marina Las Condes Hotel)**
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	**O-115, Lianlei Wang** Disruptions of *SOX9* K2 domain reduce protein stability and induce mild skeletal dysplasia and late-onset scoliosis
09:00	**O-17, Valerie Cormier-Daire** Bialelic variants in *SLC35B2* cause a novel chondrodysplasia with hypomyelinating leukodystrophy
09:15	**O-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	**O-114, Annette Madison** Clinical and molecular characterization of Chilean patients with X-linked hypophosphatemia
09:45	**O-61, Gretl Hendrickx** Heterozygous pathogenic variants in the *CBFB* gene cause a novel phenotype resembling cleidocranial dysplasia
10:00 - 10:30	**O-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	**O-02, V. Reid Sutton** Quantitative phenotypic analyses of Robinow syndrome: WNT signalling perturbation and phenotypic similarities and variability
11:15	**O-16 Nathalia Liberatoscioli Menezes de Andrade** High prevalence of growth plate related genes in a cohort of children with isolated short stature
11:30	**O-25 Sarah Smithson** Rare genetic mechanisms in skeletal dysplasia solved by reanalysis of whole exome/genome data
11:45	**O-82 Lorenzo Botto** More than meets the eye: unsuspected systemic diseases identified through skeletal dysplasias.
12:00	**O-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**
12:45 - 13:45	Lunch
13:45 - 14:05	**BioMarin corporate symposium** International Achondroplasia Management Guidance - Julie Hoover-Fong
14:05 - 15:20	**Session 3. Focus on molecular diagnosis 1 |** Moderators: Karen Heath - Geert Mortier
14:05	**O-68, Daniel Carvalho** TAR syndrome cohort of 20 Brazilian patients: variable clinical phenotype and molecular characterization of *RBM8A* variants.
14:20	**O-76, Yiming Wang** Development of a Genetic Diagnostic Algorithm for Individuals with Split Hand Foot Malformation
14:35	**O-130, Carlos Bacino** Increasing the Diagnosis Rate in Rare Bone disorders. Our Experience in the Undiagnosed Disease Network at Baylor College of Medicine
14:50	**O-14, Alistair Calder** Eiken dysplasia with Parathyroid Hormone resistance: two new paediatric cases with the same novel variant
15:05	**O-41, Bruno Marcarini** Next-generation sequencing applied to skeletal disorders: a ten-year experience in a Tertiary Center in Brazil
15:20 - 15:40	Coffee break
15:40 - 16:40	**Poster session 1 (P-05 to P-73) - Chaitén Foyer**
17:00	**Social program - free evening**

Local time GMT-4	**Friday 26th August, 2022**
08:30 - 09:45	**Session 4. Clinical series and observations |** Moderators: Alistair Calder - Ravi Savarirayan
08:30	**O-09, Sheela Nampoothiri** Skeletal Dysplasia Registry from a Tertiary Hospital from South India : 16 Years Experience
08:45	**O-35, Isabela Pasa** Molecular analysis of severe cases of Osteogenesis imperfecta and other rare low-mineral density disorders
09:00	**O-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	**O-13, Caroline Michot** Nine new cases of spondylometaphyseal dysplasia with corner fractures: enhancement of the phenotypic spectrum of *FN1* gene mutations
09:30	**O-52, Lucy Scrimshaw** Cerebral vasculopathy in osteopathia striata with cranial sclerosis, a rare but important complication.
09:45 - 10:15	Coffee break
10:15 - 11:00	**Session 5. Skeletal dysplasias: prenatal aspects |** Moderators: Melita Irving - Denise Cavalcanti
10:15	**O-43, Rhoda Akilapa** Observational study of fetal foramen magnum, thoracolumbar kyphosis and hippocampal development in Achondroplasia
10:30	**O-62, Marwan Shinawi** Diagnostic challenges of fetal skeletal dysplasia: A cohort of patients with prenatal diagnoses of skeletal dysplasias with unique or atypical postnatal courses
10:45	**O-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
11:00 - 11:30	**Lecture on Nosology, Professor Andrea Superti-Furga**
11:30 - 12:30	**ISDS Business Meeting**
12:30 - 13:30	Lunch
12:50 - 13:10	**Takeda corporate lunch symposium** Skeletal involvement in the mucopolysaccharidosis - Norberto Guelbert
13:30 - 14:30	**Session 6. Evaluation of growth, quality of life, pain and functionality |** Moderators: Roberto Mendoza - Jacqueline Hecht
13:30	**O-54, Antonio Rossi** Identification of potential non-invasive biomarkers in diastrophic dysplasia
13:45	**O-42, Penelope Ireland** Development of the Screening Tool for Everyday Mobility and Symptoms (STEMS) for Skeletal Dysplasia
14:00	**O-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)
14:15	**O-105, Natassja Billich** Nutritional issues in skeletal dysplasia: A scoping review
14:30 - 15:00	Coffee break
15:00 - 16:15	**Session 7. Biomarkers and mouse models |** Moderators: Matthew Warman - Caroline Michot
15:00	**O-75, Jacqueline Hecht** Nutraceutical treatment strategies in a mouse model of pseudoachondroplasia
15:15	**O-48, Elvire Gouze** Lentiviral vector-based gene therapy for type II collagen disorders
15:30	**O-64, Pavel Krejci** Modelling achondroplasia in mouse to address the future therapy targets
15:45	**O-95, Laurence Legeai-Mallet** Hypochondroplasia gain-of-function mutation in *FGFR3* causes defective bone mineralization in mice
16:00	**O-20, Carl Dambkowski** Low-dose infigratinib, an oral selective fibroblast growth factor receptor tyrosine kinase inhibitor, demonstrates activity in a preclinical model of hypochondroplasia
16:15 - 17:15	**Poster session 2 (P-79 to P-131) - Lounge Chaitén Foyer**
18:30 - 23:00	**Conference dinner**
20:15	**Marcteaux Award**

Local time GMT-4	**Saturday 27th August, 2022**
08:30 - 09:30	**Patients - Families - Experts - Coffee**
09:45 - 11:15	**Session 8. Clinical and observational studies |** Moderators: Debora Bertola - Lorenzo Botto
09:45	**O-107, Thais Cuperman** Quantifying Outcomes in Fibrodysplasia Ossificans Progressiva (FOP) by Patient Age: Results from an International Burden of Illness Survey
10:00	**O-39, Melita Irving** ACHieve Study: Baseline Characteristics of a Multicenter Observational Study of Children with Achondroplasia
10:15	**O-10, David Tunkel** Otolaryngology Utilization in Patients with Achondroplasia: Results from the CLARITY Study
10:30	**O-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	**O-33, Ravi Savarirayan** Medical history of children enrolled in PROPEL: A prospective clinical assessment study in children with achondroplasia
11:00	**O-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** Diagnosis and challenges of ultra-rare bone diseases: A focus on FOP - Patricia Delai
11:35 - 12:00	Coffee break
12:00 - 12:45	**Session 9. Treatment Trials for Achondroplasia |** Moderators: Julie Hoover-Fong - Reid Sutton
12:00	**O-49, Nadia Merchant** A Prospective Clinical Trial of Vosoritide in Hypochondroplasia: Baseline Demographics and Preliminary Results
12:15	**O-69, Ravi Savarirayan** A Randomized Controlled Trial of Vosoritide in Infants and Toddlers With Achondroplasia
12:30	**O-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

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