

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 -	
08:45	ISDS-115, Lianlei Wang (To be confirmed) 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 CFBF gene cause a novel phenotype resembling cleidocranial dysplasia	
10:00 - 10:30	ISDS126, 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 - 12:30	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-80, Nitika Langeh (To be confirmed) MutLands, a new advancement in the anatomic pathology of the human genome and in variant interpretation	
11:30	ISDS-16, Nathalia Liberatoscioli Menezes de Andrade High prevalence of growth plate related genes in a cohort of children with isolated short stature	
11:45	ISDS-25, Sarah Smithson Rare genetic mechanisms in skeletal dysplasia solved by reanalysis of whole exome/genome data	
12:00	ISDS-82, Lorenzo Botto More than meets the eye: unsuspected systemic diseases identified through skeletal dysplasias.	
12:15	ISDS-111, Andreas Zankl Integrating the ISDS Nosology with the G2P Project and the MONDO Disease Ontology	
12:30 - 13:00	David Rimoin lecture, Matt 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 RBMBA 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 - 10:15	Session 4: clinical series and observations Moderators: Ali Calder - Ravi Savarirayan	
08:30	ISDS-09, Sheila Nampoothiri Skeletal Dysplasia Registry from a Tertiary Hospital from South India : 16 Years Experience	
08:45	ISDS-57, Shubha Phadke (To be confirmed) Genotypic Landscape of Osteogenesis Imperfecta from India	
09:00	ISDS-35, Isabela Pasa Molecular analysis of severe cases of Osteogenesis imperfecta and other rare low-mineral density disorders	
09:15	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	
09:30	ISDS-53, Cathleen Raggio (To be confirmed) Should osteogenesis imperfecta be labeled as a low bone mass condition?	
09:45	ISDS-13, Caroline Michot Nine new cases of spondylometaphyseal dysplasia with corner fractures: enhancement of the phenotypic spectrum of FN1 gene mutations	
10:00	ISDS-52, Lucy Scrimshaw Cerebral vasculopathy in osteopathia striata with cranial sclerosis, a rare but important complication.	
10:15 - 10:45	Coffee break	
10:45 - 11:30	Session 5: skeletal dysplasias: prenatal aspects Moderators: Sheila Unger - 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 Of 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	
14:00 - 15:15	Session 6: Evaluation of growth, quality of life, pain and functionality Moderators: Roberto Mendoza -	
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 (To be confirmed) 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: Matt Warman -	
15:45	ISDS-75, Jacqueline Hecht (To be confirmed) Nutraceutical treatment strategies in a mouse model of pseudoachondroplasia	
16:00	ISDS-48, Elvire Guze 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 infliximab, 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 (Prof Gen Nishimura; Prof. Sheila Unger)	
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, Ilan Arsof Burden of Illness Survey - Quantifying Outcomes in Fibrodysplasia Ossificans Progressiva (FOP) by Patient Age: Results from an International	
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	ISDS-58, Michael Kai Tsun To Lower limb deformity correction in paediatric patients with pseudoachondroplasia: a single center experience	
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-49, 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	