POSTER SESSION

ISDS MEETING 2022

Thursday 25th August, 2022 | CHAITÉN FOYER

Local time GMT-4

15:40 - 16:40 **Session 1 (P-05 - P-73)**

P-05 Persistent growth in children with achondroplasia treated with vosoritide for two years: further evidence supporting the first precision therapy for this condition

Ravi Savarirayan, MB, BS, MD1*, Louise Tofts, MB, BS2, Melita Irving, MB, BS, MD3, William Wilcox, MD, PhD4, Carlos A. Bacino, MD5, Julie Hoover-Fong, MD, PhD6, Rosendo Ullot Font, MD7, Paul Harmatz, MD8, Frank Rutsch, MD9, Michael B. Bober, MD, PhD10, Lynda E. Polgreen, MD, MS11, Ignacio Ginebreda, MD12, Klaus Mohnike, MD13, Joel Charrow, MD14, Daniel Hoernschemeyer, MD15, Keiichi Ozono, MD, PhD16, Yasemin Alanay, MD, PhD17, Paul Arundel, MB, BS18, Shoji Kagami, MD19, Natsuo Yasui, MD19, Klane White, MD, MSc20, Howard M. Saal, MD21, Antonio Leiva-Gea, MD22, Felipe Luna-González, MD22, Hiroshi Mochizuki, MD23, Donald Basel, MB, ChB24, Dania M. Porco, MS, MBA25, Kala Jayaram, MD25, Elena Fisheleva, MD26, Lynn Han, PhD25, Jonathan Day, MB, BS, PhD26

1Murdoch Children's Research Institute, Royal Children's Hospital, and University of Melbourne, Parkville, Victoria, Australia 2Kids Rehab, The Children's Hospital at Westmead, Westmead, New South Wales, Australia 3Guy's and St. Thomas' NHS Foundation Trust, Eveli

P-07 Exploring the family experience of children aged 2-4 years receiving a daily subcu- taneous injection

Tessa Dentry (1), Jenny O'Neill (2), Kaya Gardiner (3, 4), Chloe Stutterd (1), Supriya Raj (1) & Ravi Savarirayan (1)

(1) Skeletal Therapies, Murdoch Children's Research Institute, Parkville, Australia. (2) Nursing Research, The Royal Children's Hospital, Parkville, Australia. (3) Research Operations, The Royal Children's Hospital, Parkville, Australia. (4) Infectious Diseases Group, Murdoch Children's Research Institute, Parkville, Australia

P-15 How Sedaghatian-like is "Spondylometaphyseal Dysplasia Sedaghatian-like"? A comparison of recent cases

Alistair D Calder (1), Niha Peshimam (2), Hani Farah (2), Richard Caswell (3), Sian Ellard (3), Wajanat Jan (4,5), Jan Cobben (4,6) Ujwal Kariholu (2,4) Harry G. Leitch (4,6,7,8)

(1) Radiology, Great Ormond Street Hospital for Children NHS Foundation Trust, London WC1N 3JH, UK. (2) Neonatal Unit, Imperial College Healthcare NHS Trust, London, UK. (3) Exeter Genomics Laboratory, Royal Devon and Exeter NHS Foundation Trust, Exeter EX2 5DW, UK. (4) Department of Imaging, Imperial College Healthcare NHS Trust, London, UK (5) Centre for Paediatrics and Child Health, Faculty of Medicine, Imperial College London, London W2 1PG, UK. (6) North West Thames Genetics Service, Northwick Park and St. Mark's Hospitals, London, HA1 3UJ, UK. (7) Institute of Clinical Sciences, Faculty of Medicine, Imperial College London, London W12 0NN, UK. (8) MRC London Institute of Medical Sciences, London, UK

P-18 Qualitative research in children and parents of children with achondroplasia: medical challenges and impacts and their assessment using patient-reported outcome measures

Susan D. Mathias (1), Julie Hoover-Fong (2), Ravi Savarirayan (3), Chandler Crews (4), Inês Alves (5), Susana Noval (6), Amer Haider (7), Terry Cho (8), Anne Lee (9), Hilary H. Colwell (1), Elena Muslimova (9)

(1) Health Outcomes, Health Outcomes Solutions, Winter Park, United States of America; (2) Genetic Medi- cine and Pediatrics, Johns Hopkins University, Baltimore, United States of America; (3) Genetics Services, Murdoch Children's Research Institute and University of Melbourne, Melbourne, Australia; (4) Achondroplasia, The Chandler Project, Little Rock, United States of America; (5) Skeletal Dysplasia, ANDO Portugal, Évora, Portugal; (6) Achondroplasia, Fundación ALPE Acondroplasia, Oviedo, Spain; (7) Business Development, The University of Illinois at Urbana Champaign, Champaign, United States of America; (8) Clinical Operations, QED Therapeutics, Inc., San Francisco, United States of America; (9) Clinical Development, QED Therapeutics, Inc., San Francisco, United States of America

P-22 Human Metaphyseal Dysplasias Resulting from Mutations in *ANKH, COL10A1, FLNA, LRRK1, MMP9, MMP13, PLEKHM1, PTH1R, SFRP4* and *SNX10*

Robert Brommage (1)

(1) BoneGenomics, The Woodlands, Texas, USA

P-23 Novel homozygous nonsense *HES7* variant causing spondylocostal dysostosis and distal vaginal atresia

Daniel Rocha de Carvalho ( ), Janime Barboza Rangel (1), Cynthia Costa e Silva (2), Savana Camila de Lima Santos (2), Cicero Ricardo Gomes (3)

(1) Medical Residency in Medical Genetics, Health Department of Federal District, Brasilia-DF, Brazil; (2) Molecular Biology Laboratory, SARAH Network of Rehabilitation Hospital, Brasilia-DF, Brazil; (3) Department of Orthopedics, SARAH Network of Rehabilitation Hospital, Brasilia-DF, Brazil

P-24 Defining the phenotypic spectrum of cases with biallelic *PRKG2* variants and their pathogenic mechanism

Francisca Díaz-González (1,2), Yasemin Alanay (3), N Gönenli-Mungan (4), Serdar Ceylaner (5), Manuel Parrón-Pajares (2,6), Gen Nishimura (7), Karen E. Heath (1,2,8)

(1) Institute of Medical & Molecular Genetics (INGEMM), Hospital Universitario La Paz, Universidad Autónoma de Madrid, IdiPAZ, Madrid, Spain. (2) Multidisciplinary Skeletal dysplasia (UMDE-ERN BOND), Hospital Universitario La Paz, Madrid, Spain. (3) Pediatric Genetics Unit, Department of Pediatrics, Acibadem University, Istanbul, Turkey. (4) Pediatric Endocrinology and Metabolism Unit, Department of Pediatrics, Cukurova University, Adana Turkey. (5) Medical Genetics, Intergen Laboratory, Ankara, Turkey. (6) Dept of Radiology, Hospital Universitario La Paz, Madrid, Spain. (7) Center for Intractable Disease, Saitama Medical University Hospital, Saitama, Japan. (8) CIBERER, ISCIII, Madrid, Spain

P-26 Evolution of phenotype in Somali children with Cranioectodermal dysplasia associated with a novel *WDR35* variant

Sarah Smithson (1), Christine Burren (2), James Turner (3), Amaka Offiah (4), Christine Hall (5), Emma Baple (6), Maggie Williams (6)

(1) Department of Clinical Genetics, University Hospitals Bristol and Weston NHS Trust, Bristol, UK, (2) Department of Paediatric Endocrinology and Diabetes, Bristol Royal Hospital for Children, University Hospitals Bristol and Weston NHS Trust, Bristol, UK, (3) Department of Paediatric Orthopaedics, Bristol Royal Hospital for Children, University Hospitals Bristol and Weston NHS Trust, Bristol, UK., (4) Department of Oncology and Metabolism, University of Sheffield, Sheffield, UK, (5) Emeritus, Department of Paediatric Radiology, Great Ormond Street Hospital, London, UK, (6) Bristol Genetics Laboratory, South-West Genomics Laboratory, North Bristol NHS Trust, Bristol, UK

P-27 Four cases of Spondyloenchondrodysplasia with and without immune manifestations

Jimena Dri (1) , María Jose Guillamondegui (2), Eugenia Dos Santos(1), Marcela Pereyra (1), Celeste Ballester (3), Ana Tolin(3), Cristina Gatica (1)

(1) Servicio de Crecimiento y Desarrollo. Hospital Pediátrico H.J. Notti. Mendoza, Argentina. (2) Sección de Genética Clínica. Hospital Pediátrico H.J. Notti. Mendoza, Argentina. (3) Servicio de Inmunología y Alergias. Hospital Pediátrico H.J. Notti. Mendoza, Argentina

P-29 *KDELR2*-related osteogenesis imperfecta: clinico-radiological and genetic features in two families with compound heterozygous and homozygous deletions

Atif Sabir (1,2), Catherine DeVile (3), Jeremy Allgrove (4), Belinda Crowe (3), Alistair Calder (5), Deborah Shears (6), Jing Yu (7), Alistair Pagnamenta (7), Jenny Taylor (7), Fleur van Dijk (8,9)

(1) West Midlands Regional Clinical Genetics Unit, Birmingham Women's and Children's NHS FT, Birmingham, UK. (2) Institute of Cancer and Genomics, College of Medical and Dental Sciences, University of Birmingham, UK. (3) Paediatric Neurosciences, Great Ormond Street Hospital for Children NHS Foundation Trust, London, UK. (4) Paediatric Endocrinology, Great Ormond Street Hospital for Children NHS Foundation Trust, London UK. (5) Paediatric Radiology, Great Ormond Street Hospital for Children NHS Foundation Trust, London, UK. (6) Oxford Centre for Genomic Medicine, Nuffield Orthopaedic Centre, Oxford University Hospitals NHS Foundation Trust, Oxford, UK. (7) Wellcome Centre for Human Genetics, Oxford University, Oxford, Oxfordshire, UK; NIHR Biomedical Research Centre, Oxford, UK. (8) NorthWest Thames Regional Genetics Service, London North West University Healthcare NHS Trust, Watford, UK. (9) Department of Metabolism, Digestion and Reproduction, Section of Genetics and Genomics, Imperial College London, London, UK.

P-31 Evaluation of body mass index and metabolic parameters in children with achondroplasia participating in the PROPEL study

Ravi Savarirayan (1), Josep Maria De Bergua (2), Paul Arundel (3), Jean Pierre Salles (4), Antonio Leiva- Gea (5), Melita Irving (6), Vrinda Saraff (7), Helen McDevitt (8), Fernando Santos-Simarro (9), Marc Nicolino (10), Valerie Cormier-Daire (11), Peter Kannu (12), Mars Skae (13), Michael B. Bober (14), John Phillips III (15), Christine Burren (16), Paul Harmatz (17), Howard Saal (18), Julie Hoover-Fong (19), Elena Muslimova (20), Terry Cho (21), Richard Weng (22), Daniela Rogoff (20)

(1) Genetics Services, Murdoch Children's Research Institute and University of Melbourne, Melbourne, Australia; (2) Orthopedics, Unidad Cirugía Artroscópica, Hospital Vithas Vitoria, Vitoria-Gasteiz, Spain; (3) Children's & Adolescent Services, Sheffield Children's NHS Foundation Trust, Sheffield, United Kingdom; (4) Endocrine, Bone Diseases, and Genetics Unit, Children's Hospital, Toulouse University Hospital, Toulouse, France; (5) Pediatric Endocrinology, Instituto de Investigación Biomédica de Málaga and Hospital Universitario Virgen de la Victoria, Málaga, Spain; (6) Clinical Genetics, Guy's and St Thomas' NHS Trust, London, United Kingdom; (7) Paediatric Endocrinology and Diabetes, Birmingham Women's and Children's NHS Foundation Trust, Birmingham, United Kingdom; (8) Neonatal and Children's Services, NHS Greater Glasgow and Clyde, Glasgow, United Kingdom; (9) Clinical Genetics, Instituto de Genética Médica y Molecular, Hospital Universitario La Paz, Madrid, Spain; (10) Paediatric-Endocrinology Unit, Hôpital Femme Mère Enfant, Hospices Civils de Lyon, Lyon, France; (11) Genetics, Imagine Institute, Hôpital Necker-Enfants Malades, University of Paris, Paris, France; (12) Medical Genetics, University of Alberta, Edmonton, Canada; (13) Paediatric Endocrinology, Manchester University NHS Foundation Trust, University of Manchester, Manchester, United Kingdom; (14) Pediatrics, Nemours Children's Hospital, Wilmington, United States of America; (15) Pediatric Medical Genetics, Vanderbilt University Medical Center, Nashville, United States of America; (16) Paediatric Endocrinology, University Hospitals Bristol and Weston NHS Foundation Trust, Bristol, United Kingdom; (17) Gastroenterology and Nutrition, University of California, San Francisco, Benioff Children's Hospital Oakland, Oakland, United States of America; (18) Pediatrics, Cincinnati Children's Hospital Medical Center, Cincinnati, United States of America; (19) Genetic Medicine and Pediatrics, Johns Hopkins University, Baltimore, United States of America; (20) Clinical Development, QED Therapeutics, Inc., San Francisco, United States of America; (21) Clinical Operations, QED Therapeutics, Inc., San Francisco, United States of America; (22) Biostatistics, QED Therapeutics, Inc., San Francisco, United States of America

P-32 Infigratinib in children with achondroplasia: design of the PROPEL, PROPEL2 and PROPEL OLE studies

Ravi Savarirayan (1), Josep Maria De Bergua (2), Paul Arundel (3), Helen McDevitt (4), Valerie Cormier- Daire (5), Vrinda Saraff (6), Mars Skae (7), Borja Delgado (8), Antonio Leiva-Gea (9), Fernando Santos- Simarro (10), Jean Pierre Salles (11), Marc Nicolino (12), Massimiliano Rossi (13), Peter Kannu (14), Michael B. Bober (15), John Phillips III (16), Howard Saal (17), Paul Harmatz (18), Christine Burren (19), Garrett Gotway (20), Terry Cho (21), Elena Muslimova (22), Richard Weng (23), Daniela Rogoff (22), Julie Hoover-Fong (24), Melita Irving (25)

(1)GeneticsServices,MurdochChildren'sResearchInstituteandUniversityofMelbourne,Melbourne,Australia; (2) Orthopedics, Unidad Cirugía Artroscópica, Hospital Vithas Vitoria, Vitoria-Gasteiz, Spain; (3) Children's & Adolescent Services, Sheffield Children's NHS Foundation Trust, Sheffield, United Kingdom; (4) Neonatal and Children's Services, NHS Greater Glasgow and Clyde, Glasgow, United Kingdom; (5) Genetics, Imagine Institute, Hôpital Necker-Enfants Malades, University of Paris, Paris, France; (6) Paediatric Endocrinology and Diabetes, Birmingham Women's and Children's NHS Foundation Trust, Birmingham, United Kingdom; (7) Paediatric Endocrinology, Manchester University NHS Foundation Trust, University of Manchester, Manchester, United Kingdom; (8) Clinical Genetics, Hospital Universitario Virgen de la Victoria, Málaga, Spain; (9) Pediatric Endocrinology, Instituto de Investigación Biomédica de Málaga and Hospital Universitario Virgen de la Victoria, Málaga, Spain; (10) Clinical Genetics, Instituto de Genética Médica y Molecular, Hospital Universitario La Paz, Madrid, Spain; (11) Endocrine, Bone Diseases, and Genetics Unit, Children's Hospital, Toulouse University Hospital, Toulouse, France; (12) Paediatric-Endocrinology Unit, Hôpital Femme Mère Enfant, Hospices Civils de Lyon, Lyon, France; (13) Medical Genetics, Lyon University Hospital, Lyon, France; (14) Medical Genetics, University of Alberta, Edmonton, Canada; (15) Pediatrics, Nemours Children's Hospital, Wilmington, United States of America; (16) Pediatric Medical Genetics, Vanderbilt University Medical Center, Nashville, United States of America; (17) Pediatrics, Cincinnati Children's Hospital Medical Center, Cincinnati, United States of America; (18) Gastroenterology and Nutrition, University of California, San Francisco, Benioff Children's Hospital Oakland, Oakland, United States of America; (19) Paediatric Endocrinology, University Hospitals Bristol and Weston NHS Foundation Trust, Bristol, United Kingdom; (20) Pediatrics - Genetics and Metabolism, Children's Medical Center Dallas, Dallas, United States of America; (21) Clinical Operations, QED Therapeutics, Inc., San Francisco, United States of America; (22) Clinical Development, QED Therapeutics, Inc., San Francisco, United States of America; (23) Biostatistics, QED Therapeutics, Inc., San Francisco, United States of America

P-36 A systematic review of the effectiveness and safety of therapeutic options for patients with achondroplasia and clinical unmet need

Smith, A (1), Musat, M (2), Harricharan, S (2), Vurgun, N (2), Noori, W (1)

(1) Ascendis Pharma Inc, Palo Alto, USA (2) Cytel, Inc., Waltham, USA

P-37 A systematic review on the clinical burden of achondroplasia and management of achondroplasia-associated complications

Noori, W (1), Musat, M (2), Harricharan, S (2), Vurgun, N (2), Smith, A (1)

(1) Ascendis Pharma Inc, Palo Alto, USA (2) Cytel, Inc., Waltham, USA

P-38 Baseline Characteristics of the ACcomplisH Phase 2 Trial Evaluating Once-Weekly TransCon CNP in Children with Achondroplasia

Savarirayan, R (1,2), Abuzzahab, MJ (3), Bacino, CA (4), Bober, MB (5), Hoernschemeyer, DG (6), Hofman, PL (7), Högler, W (8) Hove, HB (9), Legare, JM (10), Ma, N (11), Quattrin, T (12), McDonnell, C (13), Schnabel, D (14), Sousa, SB (15), White, KK (16), Zarate, Y (17), Zhang, Y (18), Viuff, D (19), Harloff-Nielsen, M (19)

(1) Murdoch Children's Research Institute, Parkville, Australia (2) University of Dublin, Trinity College, Dublin, Ireland (3)Childrens Hospitals and Clinics of Minnesota, Minneapolis, USA (4) Baylor College of Medicine, Houston, USA (5) Nemours Childrens Hospital, Wilmington, USA (6) University of Missouri Women's and Children's Hospital, Columbia, USA (7) The Liggins Institute, University of Auckland, Auckland, New Zealand (8) Johannes Kepler University, Linz, Austria (9) Copenhagen University Hospital Rigshospitalet, Copenhagen, Denmark (10) University of Wisconsin School of Medicine and Public Health, Madison, USA (11) Children's Hospital Colorado, Aurora, USA (12) University at Buffalo, Buffalo, USA (13) Children's Health Ireland at Temple Street, Dublin, Ireland (14) Center for Chronically Sick Children, Charité – University Medicine Berlin, Berlin, Germany (15) Hospital Pediátrico de Coimbra, Coimbra, Portugal (16) Seattle Children's Hospital, Seattle, USA (17) University of Arkansas for Medical Sciences, Little Rock, USA (18) Ascendis Pharma Inc., Palo Alto, USA (19) Ascendis Pharma A/S, Hellerup, Denmark

P-40 IPSC reprogramming to create the first SEMD (biglycan type) iPSC-derived chondrocyte models

De Kinderen P. (1), Peeters S. (1), Mortier G. (3), Loeys B. (1,2), Woods S. (4), Verstraeten A. (1), Meester J. (1)

(1) Center of Medical Genetics, Faculty of Medicine and Health Sciences, University of Antwerp and Antwerp University Hospital, Antwerp, Belgium . (2) Department of Human Genetics, Radboud University Nijmegen Medical Center, Nijmegen, The Netherlands. (3) Center of Human Genetics, University of Leuven and Leuven University Hospital, Leuven, Belgium. (4) Division of Cell Matrix Biology and Regenerative Medicine, School of Biological Sciences, Faculty of Biology, Medicine and Health, University of Manchester, Manchester, UK

P-46 A girl with cutaneous skeletal hypophosphatemia syndrome due to somatic *RAS* mutations

Mariana del Pino (1)Gisela L. Viterbo (2); Silvina Abbate (3), Josefina Chinton (4), Andrea Bettina Cervini (5); María Gabriela Obregón (3) and Virginia Fano (1)

(1)Growth and development. Hospital Garrahan. Buenos Aires. Argentina (2) Endocrinology. Hospital Garrahan. Buenos Aires. Argentina (3) Genetics. Hospital Garrahan. Buenos Aires. Argentina (4) Molecular Laboratory. Genetics. Hospital Garrahan. Buenos Aires. Argentina(5) Dermatology. Hospital Garrahan. Buenos Aires. Argentina

P-47 WIDE PHENOTYPIC VARIABILITY IN ACAN DELETIONS: DESCRIPTION OF FOUR FAMILIES

Sentchordi-Montané L (1,2), Díaz-Gonzalez F (2), Modamio-Høybjør S (2), Carcavilla-Urquí A (2,3), Saraiva J (4,5), Kant S (6), de Bruin C (7), van_Duyvenvoorde H.A. (7), González Cabaleiro I (8), Rey Cordo C.L. (8), Chamorro Martín J.L. (8), Sousa S.B. (4,5), Heath K.E. (2,9)

(1) Hospital Universitario Infanta Leonor, Madrid. (2) Institute of Medical and Molecular Genetics (INGEMM), IdiPAZ, UAM and Skeletal dysplasia multidisciplinary Unit (UMDE), Hospital Universitario La Paz, Madrid, Spain. (3) Pediatric Endocrinology, Hospital Universitario La Paz, Madrid, Spain. (4) Medical Genetics Unit, Hospital Pediátrico, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal. (5) University Clinic of Genetics, Faculty of Medicine, Universidade de Coimbra, Coimbra, Portugal. (6) Department of Clinical Genetics. Erasmus MC, University Medical Center Rotterdam, The Netherlands. (7) Laboratory for Diagnostic Genome analysis (LDGA), Dept. of Clinical Genetics, LUMC, Leiden, The Netherlands. (8) Hospital Universitario de Vigo, Spain. (9) CIBERER, ISCIII, Madrid, Spain

P-50 Novel candidate regions associated with Femoral Facial Syndrome

Maria Dora Lacarrubba-Flores (5),(Yanca Gasparini(1), Leslie D. Kulikowski (2), Daniel Rocha Carvalho (3), Erlane Marques Ribeiro (4), Denise Pontes Cavalcanti (5)

(1) Cytogenomics Laboratory, Department of Pathology, and Genetics Unit, Department of Pediatrics, Instituto da Criança, Hospital das Clinicas, Faculty of Medicine, University of São Paulo, São Paulo, Brazil (2) Cytogenomics Laboratory, Department of Pathology, Faculty of Medicine, University of São Paulo, São Paulo,, Brazil (3) Genetic Unit, SARAH Network of Rehabilitation Hospital, Federal District, Brasília, Brazil. (4) Department of Pediatrics, Children´s Hospital Albert Sabin, Fortaleza, Ceará, Brazil. (5) Department of Translational Medicine, Faculty of Medical Sciences, University of Campinas, Brazil

P-51 Adaptation of skeletal dysplasia patient treatment guidelines to condensed, rapid-access checklists and video vignettes for patients and healthcare provider

Julie Hoover-Fong, MD, PhD (1), Wagner AR Baratela, MD, PhD (2), Michael Bober, MD, PhD (3), Viviana Bompadre, PhD (4), Moira Cheung, MD, PhD (5), Tae-Joon Cho, MD, PhD (6), Michael Goldberg, MD * (7), Melita Irving, MD (8), Shawn Kamps, MD (9), William Mackenzie, MD* (10), Cathleen Raggio, MD (11), Ravi Savarirayan, MBBS* (12), Samantha Spencer, MD (13), Klane White, MD* (4)

(1) Department of Genetic Medicine, Johns Hopkins University, Baltimore, USA (2) Department of Pediatrics, Santa Casa de Misericórdia School of Medical Sciences, São Paulo, Brazil (3) Department of Pediatric Genetics, Nemours Children's Hospital, Wilmington, USA (4) Department of Orthopedics and Sports Medicine, University of Washington, Seattle Childrens Hospital, Seattle, USA (5) Department of Pediatrics, Guy's and St Thomas' NHS Foundation Trust, London, UK (6) Department of Orthopedics, Seoul National University Children's Hospital, Seoul, South Korea (7) Schwartz Center for Compassionate Healthcare, Boston Children's Hospital, Boston, USA (8) Department of Genetic Medicine, Guy's and St Thomas' NHS Foundation Trust, London, UK (9) Department of Radiology, University of Washington, Seattle Childrens Hospital, Seattle, USA (10) Department of Pediatric Orthopedics, Nemours Children's Hospital, Wilmington, USA (11) Department of Pediatric Orthopedics, Hospital for Special Surgeries, New York, USA (12) Victorian Clinical Genetics Services, Murdoch Childrens Research Institute, Melbourne, Australia (13) Department of Orthopedic Surgery, Boston Children's Hospital, Boston, USA *Executive Founding Commitee

P-59 Prenatal genetic testing can lead to (almost) self-fulfilling prophecy: diastrophic dysplasia as an example

Andrea J Schelhaas (1), Angela L Duker (1), Michael B Bober (1, 2), Ricki S Carroll (1,2)

(1) Nemours Children's Hospital, Wilmington, DE, USA. (2) Thomas Jefferson University, Philadelphia, PA, USA

P-60 Identification of a homozygous missense variant in *SP7*/Osterix in a new form of craniodiaphyseal dysplasia

Grett Hendrickx (1, 2), Eveline Boudin (1), Ellen Steenackers (1), Corinne Collet (3), Geert R. Mortier (1, 2), David Geneviève (4), Wim Van Hul (1)

1) Department of Medical Genetics, Antwerp University Hospital and University of Antwerp, 2650 Edegem, Belgium. (2) Laboratory for Skeletal Dysplasia Research, Department of Human Genetics, KU Leuven, Herestraat 49, 3000, Leuven, Belgium (3) INSERM U1132 and Université de Paris, Reference Centre for Rare Bone Diseases, Hospital Robert Debré, F-75010 Paris, France (4) INSERM U1183 and Montpellier University, Reference Center for Bone Diseases, Clinical Genetics Unit, Montpellier University Hospital, Montpellier, France

P-63 Expanding clinical and molecular evidence of *CYP26B1* involvement in retinoic acid catabolism defects

Karina da Costa Silveira (1), Inara Chacon Fonseca (2), Connor Oborn (1), Saima Ghafoor (1), Oswaldo Artigalas (3), Aline Iacovone (4), Eric Campos (5,6), Denise Pontes Cavalcanti (4), Peter Kannu (1)

(1) Department of Medical Genetics, University of Alberta, Edmonton, AB, Canada (2) Division of Clinical and Metabolic Genetics, Department of Pediatrics, The Hospital for Sick Children, University of Toronto, Toronto, ON, Canada (3) Clinical Genetics Unit, Children's Hospital, Grupo Hospitalar Conceição, Porto Alegre, RS, Brazil (4) Skeletal Dysplasia Group, University of Campinas, Campinas, SP, Brazil (5) Genetics & Genome Biology program, Hospital for Sick Children, Toronto, ON, Canada (6) Department of Molecular Genetics, University of Toronto, Toronto, ON, Canada

P-67 Biallelic loss of function variants in *RFLNA* and spondylo-carpal-tarsal synostosis syndrome

B. Campos-Xavier (1), M. Gnoli (2), D. Krakow (3), S. P. Robertson (4), L. Sangiorgi (2), S. Unger (1), A. Superti- Furga (1)

(1)Div. of Genetic Medicine, Lausanne University Hospital, Lausanne, Switzerland (2) Med. Genetics Division, Istituto Ortopedico Rizzoli, Bologna, Italy (3) Depts. of Human Genetics, Pediatrics, Orthop. Surgery and Ob/ Gyn, David Geffen School of Medicine at UCLA Los Angeles, USA (4) Paediatric Genetics, Dept of Women's and Children's Health (Dunedin), University of Otago, Otago, New Zealand

P-72 Unexpected findings demonstrate the necessity for long-term follow-up in rare skeletal diseases and the importance of multidisciplinary teams

Ramos Mejía R. (1), del Pino M. (1), Huckstadt V. (2), Caino S. (1), Obregon M.G. (2), Moosa S. (3), Heath K.E. (4), Fano V. (1)

(1) Growth and Development Department, Hospital Garrahan, Buenos Aires, Argentina. (2) Genetics Department, Hospital Garrahan, Buenos Aires, Argentina. (3) Genetics Department, Stellenbosch University, Stellenbosch, South Africa. (4) Institute of Medical and Molecular Genetics (INGEMM), Madrid, España and Skeletal dysplasia multidisciplinary Unit (UMDE), Hospital Universitario la Paz, UAM, IdiPAZ, Madrid, España

P-73 ROCKET: International prospective natural history study in children with a type II collagen disorder with short stature

Samantha Parker (1), Elvire Gouze (1), Wendy Guillouche (1), Valerie Cormier-Daire (2), Julie Hoover-Fong (3), Melita Irving (4), Roberto Mendoza (5), Maria Salcedo Montejo (6), Reid Sutton (7), Sheila Unger (8), Andrea Superti-Furga (8)

(1) Innoskel, Biot, France (2) Hôpital Necker - Enfants Malades, Paris, France (3) Johns Hopkins University School of Medicine, Baltimore, Maryland USA (4) Guys and St Thomas NHS Foundation Trust, London, United Kingdom (5) University of Toronto The Hospital for Sick Children (SickKids), Toronto, Ontario Canada (6) La Paz University Hospital, Madrid, Spain (7) Baylor College of Medicine, Houston, Texas USA (8) Centre Hospitalier Universitaire Vaudois, Lausanne University Hospital, Lausanne, Switzerland

Friday 26th August, 2022 | LOUNGE CHAITÉN FOYER

Local time GMT-4

16:15 - 17:15 **Session 2 (P-79 - P-131)**

P-79 An unexpected result: another piece in the *IFITM5* puzzle?

Helen Livesey (1), Philip Dean (2), Jennifer Simmonds (2), Amaka Offiah (3), Jennifer Campbell (1)

(1) Department of Clinical Genetics, Leeds Teaching Hospitals NHS Trust, Leeds (2) North East and Yorkshire Genomics Laboratory Hub Central Lab, Leeds (3) University of Sheffield, Department of Oncology & Metabolism, Sheffield

P-83 Rescuing the molecular diagnosis of Osteochondrodysplasias in Brazil from the National Rare Genomes Project

Carolina Araujo Moreno (1,8), Angelina Xavier Acosta (2), Joanna Goes Castro Meira (3), Mariana Paes Leme Ferriani (4), Ester Silveira Ramos (5), Chong Ae Kim (6), Débora Romeo Bertola (6), Mara Lúcia Schmitz Ferreira Santos (7), Rare Genomes Project Consortium (1), João Bosco Oliveira (1), Denise Pontes Cavalcanti (8)

(1) Molecular Genetics Service, Department of Laboratory Medicine, Hospital Israelita Albert Einstein, São Paulo, Brazil. (2) Medical Genetics Service, Department of Pediatrics, Hospital Universitário Professor Edgard Santos, Federal University of Bahia, Salvador, Brazil. (3) Division of Medical Genetics, Hospital Universitário Professor Edgard Santos, Federal University of Bahia, Salvador, Brazil. (4) Departamento de Clínica Médica, Serviço de Alergia e Imunologia, Faculdade de Medicina de Ribeirão Preto, Universidade de São Paulo, Ribeirão Preto, Brazil. (5) Departamento de Genética, Faculdade de Medicina de Ribeirão Preto, Universida de São Paulo, Ribeirão Preto, Brazil. (6) Instituto da Criança, HC-FMUSP, São Paulo, Brazil. (7) Department of Pediatric Neurology, Hospital Infantil Pequeno Príncipe, Curitiba, Brazil. (8) Grupo de Displasias Esqueléticas, Área de Genética Médica, Departmento de Medicina Translacional, FCM-UNICAMP, Campinas, Brazil

P-86 Novel *BMPER* variant in a fetus with persistent notochord image on second trimester ultrasonography

Mehmet Aytac Yuksel (1), Tadahiro Mitani(2), Davut Pehlivan(2,3,4), James R. Lupski(2,3,5,6)

(1) Department of Gynecology and Obstetrics, Division of Perinatology, Acibadem University, Istanbul, Turkey (2) Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, USA (3) Texas Children's Hospital, Houston, TX, USA. (4) Division of Neurology and Developmental Neuroscience, Department of Pediatrics, BCM, Houston, TX 77030, USA (5) Department of Pediatrics, Baylor College of Medicine, Houston, TX, USA (6) Human Genome Sequencing Center, Baylor College of Medicine, Houston, Texas, 77030, USA

P-87 A new case of metaphyseal Spahr-type dysplasia due to a novel variant in *MMP13* and review of the literature

André M. Travessa (1), Silvia Modamio-Høybjør (2,3), Karen E. Heath (2,3), Ana Berta Sousa (1)

(1) Medical Genetics Department, Department of Pediatrics, and ERN-BOND, Hospital de Santa Maria, Centro Hospitalar Universitário Lisboa Norte, Lisbon, Portugal. (2) Institute of Medical and Molecular Genetics (INGEMM), Hospital Universitario La Paz, Universidad Autonóma de Madrid, IdiPAZ, and CIBERER, ISCIII, Madrid, Spain. (3) Skeletal Dysplasia Multidisciplinary Unit (UMDE) and ERN-BOND, Hospital Universitario La Paz, Madrid, Spain

P-88 IPN60130 for the Treatment of Fibrodysplasia Ossificans Progressiva: Methodology of the Randomized, Double-Blind, Placebo-Controlled Phase II FALKON Trial

Karimian N (1), Powell C (2), Shih F (2), Sleiman M (3)

(1) Ipsen, Montreal, Canada; (2) Ipsen, Cambridge, MA, US; (3) Ipsen, Boulogne-Billancourt, France

P-89 HUNGRY BONE SYNDROME ASSOCIATED TO RICKETS

Catalina Jimenez (1), Cristian Seiltgens (1)

(1) Endocrinology Unit, Department of Pediatrics, Pontificia Universidad Católica de Chile, Santiago, Chile

P-90 A Unique Variant in *COL2A1* Causes Severe Kyphoscoliosis and Moderate Short Stature

Howard Saal (1), Rachel Lombardo (1,2)

(1) Division of Human Genetics, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio, United States (2) BioMarin Pharmaceuticals, San Rafael California, United States

P-94 Disproportionate Short Stature, Limb and Skeletal Abnormalities in DEGCAGS Syndrome Caused by Pathogenic Variants in the *ZNF699* Gene.

Roberto Mendoza-Londono (1), Anna Szuto (1), Lucie Dupuis (1) Yi Wen Wang (1) and the Care for Rare Consortium

(1) Division of Clinical and Metabolic Genetics, Department of Pediatrics, The Hospital for Sick Children and University of Toronto, Canada

P-101 Legg-Calvé-Perthes disease due to missense variant in the *COL2A1* gene

Juan Garcia (1,2), Juan Barrantes (1,2), Orietta Beltran

(1,3) de Investigación BioGenÉtica & Bioderecho, Facultad de Medicina, Universidad Militar Nueva Granada, Bogotá, Colombia (2) Grupo de Interés en Genética, GENOSIG, Universidad Militar Nueva Granada, Bogotá, Colombia. Unidad de Genética (3) Organización Keralty, Bogotá, Colombia

P-103 Blomstrand dysplasia – a cluster due to a founder effect of a variant in *PTHR1* segregating over six generations in northeastern Brazil

Denise Pontes Cavalcanti (1), Luciana Cardoso Bonadia (1), Carolina Araujo Moreno (1), Isabella Monlléo (2), Mariana Novaes Santos (3), Silviene F. Oliveira (4), Kelly Nunes (5)

(1) Grupo de Displasias Esqueléticas, Área de Genética Médica, Departamento de Medicina Translacional, FCM, Universidade Estadual de Campinas (UNICAMP), Campinas, Brazil. (2) Setor de Genética Médica, Faculdade de Medicina, Hospital Universitário Prof. Alberto Antunes, Universidade Federal de Alagoas, Alagoas, Brazil. (3) Programa de Pós-graduação em Ciências Médicas, Faculdade de Medicina, Universidade Federal de Alagoas, Alagoas, Brazil. (4) Departamento de Genética e Morfologia, Universidade de Brasília, Brasília, Brazil. (5) Departamento de Genética e Biologia Evolutiva, Universidade de São Paulo, São Paulo, Brazil

P-104 A familial Venezuelan case of Strudwick type of spondyloepimetaphyseal dysplasia. Importance of the diagnostic clinical approach

Francisco Cammarata-Scalisi (1), Uta Matysiak (2), Gunda Ruzaike (2), Antonio Cárdenas Tadich (1), Maykol Araya Castillo (3), Carmen Ludeña Mendoza (1), Ana Bracho (4), Michele Callea (5)

(1) Department of Pediatrics, Regional of Antofagasta Hospital, Antofagasta, Chile (2) Department of Pediatrics, Medical Center-University of Freiburg, Faculty of Medicine, University of Freiburg, Freiburg, Germany (3) Clinical Laboratory, Regional of Antofagasta Hospital, Chile (4) Institute of Genetic Research, Faculty of Medicine, University of Zulia, Maracaibo, Venezuela (5) Pediatric Dentistry and Special Dental Care Unit, Meyer Children's University Hospital, 50139 Florence, Italy

P-106 MOLECULAR FINDINGS IN PATIENTS WITH HYPOPHOSPHATEMIC RICKETS RACHITIS IN NEUQUÉN, ARGENTINA

Silvia Avila (1,2), Mailen Costa (2), Georgina Eseni (2) Gabriela Bastida (2), Maria Freire (2), Amalia Ghiglione (2)

(1) Facultad de Ciencias Médicas Universidad Nacional del Comahue, (2) Hospital Provincial de Neuquén

P-110 Pycnodysostosis: report of a female in Chile

Michelle Zeppelin (1), Damián Gutiérrez (2), Danitza Campos (1), José Arroyo (1), Pablo Alarcón (1)

(1) Sección Genética, Hospital Clínico de la Universidad de Chile José Joaquín Aguirre, Santiago, Chile. (2) Hospital Dr. Mauricio Heyermann Torres, Angol, Chile

P-113 Short stature with advanced bone age in two Chilean families carrying mutations in *ACAN* gene

Consuelo Pino (1), Mariana Aracena (2,3), Guillermo Lay-Son (3,4), Cristián García (5)

(1) Resident Pediatric Endocrinology Unit, Pontificia Universidad Católica de Chile, Santiago, Chile. (2) Genetics Unit, Clínica Santa María, Santiago, Chile. (3) Genetics and Metabolic Diseases Unit, Pontificia Universidad Católica de Chile, Santiago, Chile. (4) Clinical Genetics Unit, Complejo Asistencial Dr. Sótero del Río. (5) Department of Radiology, Pontificia Universidad Católica de Chile, Santiago, Chile

P-116 NEW VARIANT IN *NPR2* AS A CAUSE OF SHORT STATURE AND A CROMESOMELIC DYSPLASIA IN 3 CHILEAN FAMILIES

Fernanda Peña F (1), Alejandro Martinez A.(1), Álvaro Flores (2,3), Cristián García (2), Abraham Urzúa (4), Marcela Lagos (4), Guillermo Lay-Son (5,6)

(1) Pediatric Endocrinology Unit, Pontificia Universidad Católica de Chile, Santiago, Chile; (2) Department of Radiology, Pontificia Universidad Católica de Chile, Santiago, Chile; (3) Department of Radiology, Complejo Asistencial Dr. Sótero del Río, Santiago, Chile; (4) Clinical Laboratory Department, Pontificia Universidad Católica, Santiago, Chile; (5) Clinical Genetics Unit, Pontificia Universidad Católica de Chile, Santiago, Chile; (6) Clinical Genetics Unit, Complejo Asistencial Dr. Sótero del Río, Santiago, Chile

P-117 COL1-related connective tissue overlap disorder, report of two argentinian cases

Mariana Lavia (1), Florencia Bevilacqua (1), María Jimena Díaz (1), Sofía López (1), Agostina Tardivo (1), Rosario Ramos Mejía (2), Silvina Abbate (1), Angélica Moresco (1), Virginia Fano (2)

(1) Genetics Department, J. P. Garrahan Pediatrics Hospital. Buenos Aires. Argentina. (2) Growth and Development Department, J. P. Garrahan Pediatrics Hospital. Buenos Aires. Argentina

P-120 Molecular diagnosis in a case of a fetus with angulated femurs and the provisional diagnosis of Kyphomelic Dysplasia: a contribution to the elucidation of the genotype and phenotype

Julieta De Victor (1), Guillermo Alberto(1), Josefina María Álvarez Arancedo (1), Santiago Ripodas (1), Miguel David Contreras (1), María Ester Mollica (1), Miguel Angel Aguirre (1)

(1) Centro Nacional de Genética Medica "Eduardo Castilla"

P-121 Suspected Skeletal Ciliopathy with an Inconclusive Molecular Test

D. Campos(1), J. Arroyo(1), M. Zeppelin(1), P. Alarcón(1), V. Castillo(1), R. Pardo(1,2) .

(1)Sección Genética, Hospital Clínico de la Universidad de Chile José Joaquín Aguirre, Santiago, Chile. (2)Unidad de Neonatología, Departamento de Ginecología y Obstetricia, Hospital Clínico Universidad de Chile.

P-122 Type 1 Schwartz-Jampel Syndrome: report of a case in Chile

J. Arroyo (1), D. Campos1, M. Zeppelin (1), María Morante (2), Constanza Sfeir (2), Ana María Aravena Paras (3), Daniela Avila Smirnow (4,5), María Beytía (4,5), P. Alarcón (1,3,4)

(1) Sección Genética, Hospital Clínico de la Universidad de Chile José Joaquín Aguirre, Santiago, Chile. (2) Clínica Santa María, Santiago Chile. (3) Clínica BUPA santiago, Chile. (4) Complejo Asistencial Hospital Dr Sótero del Río. (5) Red-Christus, Pontificia Universidad Católica

P-123 Suspected Renal agenesis with limb defect syndrome: report of a case in Chile

P Alarcón (1), Aurora Canessa (2), Michelle Zeppelin (1), José Arroyo (1) , Danitza Campos (1)

(1) Sección Genética, Hospital Clínico Universidad de Chile, Santiago, Chile (2) Universidad Autónoma de Chile, Talca, Chile

P-124 A suspected case of Nail-Patella syndrome in a chilean female patient

P Alarcón (1, 2)

(1) Sección Genética, Hospital Clínico Universidad de Chile, Santiago Chile. (2) Clínica BUPA, Santiago, Chile

P-127 Experience in a University Health Network's Clinical Laboratory of tests for the study of skeletal dysplasias

Marcela Lagos (1,2), Gabriel Arriata (1), Alejandra Vera (2), Abraham Urzúa (2), Cecilia Mellado (3), Mariana Aracena (3), Guillermo Lay-Son (3), Gloria Durán (3), Ligia Valdivia (2)

(1) Clinical Laboratory Department, Pontificia Universidad Católica de Chile, Santiago, Chile. (2) Molecular Biology Laboratory, Red Salud UC CHRISTUS, Santiago, Chile. (3) Clinical Genetics Unit, Pediatrics Division, Pontificia Universidad Católica de Chile Santiago, Chile

P-128 STUDY GROUP OF GENETIC SKELETAL ABNORMALITIES (GEAGE): CHILEAN EXPERIENCE IN THE DEVELOPMENT AND UNDERSTANDING OF SKELETAL DYSPLASIAS

Javiera Vildoso (1), Cecilia Mellado (1,2), Mariana Aracena (1,3), Gloria Durán (1), Marcela Lagos (4,5), Guillermo Lay-Son (1,2), Cristian García (6)

(1) Clinical Genetics Unit, Pontificia Universidad Católica de Chile, (2) Clinical Genetics Unit, Complejo Asistencial Dr. Sótero del Río, (3) Clinical Genetics Unit, Hospital Dr. Luis Calvo Mackenna, (4) Clinical Laboratory Department, Pontificia Universidad Católica, (5) Molecular Biology Laboratory, Red Salud UC CHRISTUS, (6) Department of Radiology, Pontificia Universidad Católica de Chile

P-129 Acheiropodia: Case Report

Silvia Avila (2), Estefania Ventimiglia Lagar (1), Celeste Muntaner (2)

(1) Consultorios Femina (2) Facultad de Ciencias Médicas Universidad Nacional del Comahue

P-131 A Chilean case of spondyloepimetaphyseal dysplasia and joint laxity, type 2

Mariana Aracena (1), Carolina Cares (2), Valentina Micolich (3), Cristián García(4)

(1) Unidad de Genética y Enfermedades Metabólicas, División de Pediatría, Pontificia Universidad Católica de Chile, Santiago, Chile, (2) Unidad de Genética, Hospital Dr. Luis Calvo Mackenna, Santiago, Chile, (3) Servicio Neurología, Hospital Dr. Luis Calvo Mackenna, Santiago, Chile (4) Departamento Radiología, Pontificia Universidad Católica de Chile