12th ISDS MEETING ISTANBUL 2015

July 29-August 1, 2015

Hilton İstanbul Bosphorus Hotel, Turkey

WEDNESDAY JULY 29, 2015

14.00 - 20.00	REGISTRATION
19.00 - 21.00	*WELCOME COCKTAIL*

THURSDAY JULY 30, 2015

09.15 - 09.30	WELCOME
09.30 - 10.30	*TREASURES of ISTANBUL "SİRKECİ RUINS" Speaker: Evren Türkmenoğlu*
10.30 - 11.00	**COFFEE BREAK**
11.00 - 12.30 **Chairs:**	**SESSION-I** **Andrea SUPERTI-FURGA, Beyhan TÜYSÜZ**
11.00 - 11.25	**Shiro Ikegawa** - *Combined genetics for rare and common bone and joint diseases*
11.25 - 11.50	**Karen E. Heath** - *Custom targeted NGS panel achives a high success diagnosis novel and rare skeletal dysplasias*
11.50 - 12.15	**Stefan Mundlos** - *Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions*
12.15 - 14.00	**LUNCH**
14.00 - 15.30 **Chairs:**	**SESSION-II** **Ravi SAVARIRAYAN, Luca SANGIORGI**
14.00 - 14.15	**Luisa Bonafé** - *NBAS mutations cause a multisystem disorder involving bone, connective tissue, liver, immune system and retina*
14.15 - 14.30	**David Sillence** - *A spondylo-ocular syndrome resulting from homozygous mutations in the gene encoding Xylose Transferas 2 (XYLT2)*
14.30 - 14.45	**Sheila Unger** - *Mutations in LONP1, a mitochondrial matrix protease, cause CODAS syndrome*
14.45 - 15.00	**Oliver Semler** - *Cranial ossification defects and peripheral fractures: Mutations in SEC 24 D mimicking Cole Carpenter Syndrome*
15.00 - 15.15	**Andrea Superti-Furga**-*A new form of osteolysis linked to ASAH1 (acid ceramidase) mutations*
15.15 - 15.30	**Anika Wehrle**-*ACG1A may be caused by mutations in LBR*
15.30 - 16.00	**POSTER VIEWING / COFFEE BREAK**

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16.00 - 17.15	**SESSION-III**
Chairs:	**David SILLENCE, Valerie CORMIER-DAIRE**
16.00 - 16.15	**Roberto Mendoza-Londono -** *Recessive osteogenesis imperfecta caused by mutations in SPARC*
16.15 - 16.30	**Peter Kannu -** *Mutations preventing regulated exon skipping of a receptor tyrosine kinase cause a developmental disorder of osteogenesis*
16.30 - 16.45	**Luca Sangiorgi -** *Genotype-phenotype correlation in 356 patients with dominant Osteogenesis Imperfecta*
16.45 - 17.00	**Caroline Michot**-*Application of next generation sequencing to targeted analysis of genes involved in osteogenesis imperfecta*
17.00 - 17.15	**Carlos Bacino-***Increased bone turnover, osteopenia, tibial bowing, fractures, and scoliosis in an SATB2 mutation*
17.15 - 17.30	**Igor Fijalkowski -** *LRP4 mutations in sclerosteosis and Cenani-Lenz syndrome impair sclerostin action via different mechanisms*
18.00	***DEPARTURE from HILTON HOTEL for VISIT to ISTANBUL ARCHEOLOGY MUSEUM***
20.00	***FREE NIGHT in NIŞANTAŞI DISTRICT***

FRIDAY JULY 31, 2015

09.00 - 10.30	**SCIENTIFIC SESSION-IV**
Chairs:	**Stefan MUNDLOS, Sarah NIKKEL**
09.00 - 09.15	**Andreas Zankl -** *The SKELETOME Patient Archive - A global repository of patients with skeletal dysplasias*
09.15 - 09.30	**Malte Spielmann -** *Copy number induced misregulation of developmental genes is associated with limb malformation*
09.30 - 09.45	**Sarah Smithson -** *Clinical, radiological and genetic findings in Cerebro-Costo-Mandibular syndrome*
09.45 - 10.00	**Peter Turnpenny -** *A distinctive autosomal recessive axial skeletal phenotype with Müllerian anomalies due to TBX6 gene variants*
10.00 - 10.15	**Hülya Kayserili -** *Mutations in novel gene controlling WNT-signalling cause discrete limb anomalies ranging from AARRS to Tetra-amelia*
10.15 - 10.30	**Maha Faden -** *RSPRY1 Mutations Cause a Progressive Skeletal Dysplasia*

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10.30 - 11.00	**POSTER VIEWING / COFFEE BREAK**
11.00 - 12.15 **Chairs:**	**SCIENTIFIC SESSION-V** **Stephen ROBERTSON, Sarah SMITHSON**
11.00 - 11.30	**Noriyuki Tsumaki**-Statins in Achondroplasia Treatment (Invited Speaker)
11.30 - 11.45	**Antonio Rossi -** *N-acetylcysteine treatment ameliorates the skeletal phenotype in a mouse model of Diastrophic Dysplasia*
11.45 - 12.00	**Antonella Forlino -** *Altered cytoskeleton organization affects the phenotype of Brtl mice, model for classical osteogenesis imperfecta*
12.00 - 12.15	**Michael Briggs -** *ER stress as a shared therapeutic target in genetic skeletal diseases*
12.15 - 12.45	**MEMBERS' MEETING**
12.45 - 14.00	**LUNCH**
14.00 - 15.00 **Chairs:**	**SCIENTIFIC SESSION-VI** **Luisa BONAFE, Nursel ELÇİOGLU**
14.00 - 14.15	**Ekkehart Lausch -** *Golgi or cilia? The molecular pathogenesis of TRIP11-associated disorders*
14.15 - 14.30	**Miriam Schmidts -** *Mutations in a novel dynein-2 light chain, TCTEX1D2, cause Jeune Syndrome with incomplete penetrance*
14.30 - 14.45	**Valérie Cormier-Daire -** *Molecular analysis of 24 EVC syndrome individuals using NGS and array CGH technology*
14.45 - 15.00	**Geert Mortier**-*Evidence for autosomal dominant inheritance in chondroectodermal dysplasia?*
15.00 - 15.30	**POSTER VIEWING / COFFEE BREAK**
15.30 - 16.10 **Chairs:**	**SCIENTIFIC SESSION-VII** **Jurgen SPRANGER, Zeynep YAZICI**
15.30 - 15.40	**Belinda Xavier-***Clinical and molecular characterization of 15 patients with metaphyseal enchomdromatosis.*
15.40 - 15.50	**Sergio Sousa-***Genotype and Phenotype in Lenz-Majewski syndrome*
15.50 - 16.00	**Pelin ÖŞ Kiper-***Clinical and Molecular Analysis of 3M Syndrome in a Group of Turkish Patients*
16.00 - 16.10	**Christine Hall-***Spondylo-metaphyseal acroscyphodysplasia - a second case*
16.10 - 16.20	**Alistair Calder**-*Imaging protocols in suspected skeletal dysplasia: a survey of ISDS members*
16.15 - 16.30	**Amaka Offiah-***Reconsidering something ELSE: The Value of the ISDS "SkelDys" Platform*
16.30 - 17.00	***Istanbul: From JAMES BOND to DAN BROWN*** ***Speaker: SAFFET EMRE TONGUÇ***
18.00 - 23.00	***BOSPHORUS TOUR to EMIRGAN*** ***GALA DINNER at SAKIP SABANCI MUSEUM***

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SATURDAY AUGUST 1, 2015

09.00 - 10.30	**SCIENTIFIC SESSION-VIII**
Chairs:	**Geert MORTIER, Shiro IKEGAWA**
09.00 - 09.30	**Ravi Savarirayan-*Advances in Treatment of Achondroplasia*** *(RIMOIN LECTURE)*
09.30 - 09.45	**Patrick Yap -** *Best Practices in the Evaluation and Treatment of Foramen Magnum Stenosis in Achondroplasia During Infancy*
09.45 - 10.00	**Salih Marangoz -** *Bilateral Humeral Lengthening in patients with Achondroplasia*
10.00 - 10.15	**Vladimir Kenis -** *Guided growth for deformity correction in children with skeletal dyspasias*
10.15 - 10.30	**Ahmet Alanay**- *Spinal Deformity Correction in Skeletal Dysplasias (Invited Speaker)*
10.30 - 11.00	**POSTER VIEWING / COFFEE BREAK**
11.00 - 12.00	**SCIENTIFIC SESSION-IX**
Chair:	**Reid SUTTON**
11.00 - 11.25	**Raja Padidela**-*Treatment of Hypophosphatasia (Invited Speaker)*
11.25 - 11.40	**Zlatko Sisic -** *Safety and pharmacodynamic activity of elosulfase alfa in pediatric Morquio A patients <5 years old*
11.40 - 12.10	**AWARD CEREMONY / CLOSING REMARKS**
12.30 - 14.00	LUNCH

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ISDS 2015 POSTER SESSIONS

Thursday, July 30, 2015

POSTER SESSION 1 - 15.30 - 16.00

Melita Irving	Improving Diagnostic Rates in Skeletal Dysplasia Using Whole Exome Sequencing Combined with Clinical-Radiological Phenotyping
Ozlem Atan	Next Generation Sequencing Revealed a Novel Nonsense Mutation Involving LEPRE1 in a Family
Eda Didem Kurt-Sukur	Experience of a Skeletal Dysplasia Registry in Turkey: A Five-Years Retrospective Analysis
Paulien A. Terhal	COL2A1 Related Skeletal Dysplasias, A Study on Clinical, Radiological and Molecular Data of 93 Patients
Virginia Fano	Osteogenesis Imperfecta: levels of independence and in participation in social life in adolescents
Muhammer Ozgur Cevik	Novel mutation in COL1A1 gene in a Turkish boy with osteogenesis imperfecta?
V. Reid Sutton	Multicenter Study of OI Shows That Cesarean Section Does Not Decrease Fetal Birth Fracture Rate
Selma Demir Ulusal	A Novel Mutation in COL1A1 Gene in a Pateint with Osteogenesis Imperfecta
Amal Alhashem	Pycnodysostosis with Novel Gene Mutation in a Saudi Family
Esra Kilic	A new case of pycnodysostosis with recurrent fractures and positive response to growth hormone treatment
Ayca Dilruba Aslanger	Antenatal Diagnosis of Raine Syndrome: Prenatal and Postnatal Findings
Pınar Özge Avar	Longitudinal follow up of a newborn with Dysspondyloenchondromatosis
Genevieve Baujat	Childhood Osteoporosis and Enamel Dysplasia: Expanding the Spectrum of Duplication in RUNX2
Melita Irving	Investigating the Underlying Genetically Heterogeneous Aetiology Of Cleidocranial Dysplasia

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Friday, July 31, 2015

POSTER SESSION 2 – 10:30-11:00

Fernando Santos-Simarro	Preaxial Brachydactyly PAX3 Type
Gülsen Akay Tayfun	Nager Syndrome (Preaxial Acrofacial Dysostosis): A case report
Ahmet Cevdet Ceylan	Brachytelephalangic Chondrodysplasia Punctate with Novel ARSE mutation: A Case Report
Guillermo Lay-Son	Metacarpal and metatarsal shortening associated to maternal Graves disease and neonatal hyperthyroidism
Pinar Isguven	A Case of Adams-Oliver Syndrome
Naz Guleray	Roberts/Sc Phocomelia Syndrome: A Rare Clinical Entity
Ozlem Akgün Dogan	A Novel SMARCAL1 Mutation Associated with Schimke Immunoosseous Dysplasia: A Clinical Report
Ozgur Aldemir	A Case Report: Spondyloepimetaphysial Dysplasia, Pakistani Type
Jimena Barraza-García	Two New Cases of Primordial Dwarfism Due to Homozygous POC1A Mutations
Debora Bertola	Phenotypic Spectrum in Richieri-Costa-Pereira Syndrome
Antonio Rossi	Validation of a Cant1 Knock-In and Knock-Out Mouse as Models of Desbuquois Dysplasia Type 1
Wagner Antonio da Rosa Baratela	Case Report: A 2 Yo Brazilian Girl with an Unusual Form of Rhizomelic Chondrodysplasia Punctata
Ahmet Cevdet Ceylan	Chondrodysplasia Punctata, Brachytelephalangic Type with a Novel ARSE Mutation: A Clinical Report
Mehmet Demirel	Keutel syndrome: A Rare Clinical Entity
Denise P Cavalcanti	A novel lethal skeletal dysplasia on two unrelated Brazilian fetuses
Tugba Kalayci	New Fetal Case of Blomstrand Chondrodysplasia and Review of the Literature
Alistair Calder	Platyspondylic Lethal Skeletal Dysplasia Torrance Type: Clinical and Radiological Phenotype in 2 Unrelated Long-Term Survivors
Hande Küçük Kurtulgan	Prenatal Diagnosis of Achondrogenesis Type II: A Case Report
Shwetha Ramachandrappa	SHOX Deficiency Presenting with Isolated Short Long Bones in the Second Trimester
Cecilia Mellado	Two Deletions are Responsible for the Majority of SHOX Gene Defects in Chilean Patients
Hatice Mutlu-Albayrak	Goltz (Focal Dermal Hypoplasia) Syndrome: A Case Report
Sarah M Nikkel	Diagnosing Fibrochondrogenesis the Hard Way

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POSTER SESSION 3 – 15:00-15:30

Guilherme Lopes Yamamoto	Compound Heterozygote with a Mosaic Variant: Broadening the Phenotypic Spectrum of Ciliopathies Associated with DYNC2H1
Nilay Günes	Seven Novel Mutations in Ten Turkish Patients with Ellis-Van Crevelt Syndrome
Umut Altunoglu	New Endocrine-Cerebro-Osteodysplasia (ECO) Case: Further Validation of the Phenotype with A Novel ICK Mutation
Else Merckoll	A Novel Col2A1 Mutation (p.Gly364Cys) Results in Czech Dysplasia
Anna Oschowitzer	Infantile Myofibromatosis - A Heterogeneous Disorder?
Carlos Iván Rivera-Pedroza	Second Report of Skeletal Dysplasia Due To Mutations in Pop1
Lucia Senthordi	Broadening the Clinical Spectrum of ACAN Mutations
Gözde Yeşil	Exom Sequencing Revelaed COL27A1 Mutation in a Patient with Skeletal Dysplasia without Dislocation
Patrick Yap	Pseudodiastrophic Dysplasia: Two Cases Delineating and Expanding the Pre- and Postnatal Phenotype
Pelin Ozlem Simsek-Kipe	A Novel NKX3-2 Mutation Associated with Spondylo-Megaepiphyseal-Metaphyseal Dysplasia in a Neonate: A Rare Clinical Entity
Gulen Eda Utine	Skeletal Dysplasia with Intellectual Disability: Dyggve-Melchior-Clausen Dysplasia
Saliha Yılmaz	Genotype-Phenotype Correlation in Twenty Turkish Patients with Camptodactyly-Arthropathy-Coxavara-Pericarditis Syndrome
Cormier-Daire V	Clinical and Molecular Study of a Series of 31 Patients with Chondrodysplasia with Multiple Dislocations
Esra Kilic	Progressive Pseudorheumatoid Condrodysplasia: Two New Patient with WISP3 Analysis
Franziska Friedrich	Mutations in RMRP Result in Increased HIF1A Levels
Evren Gumus	A Nonsense Mutation of the EXT1 Gene in a Turkish Patient with Multipl Cartilaginous Exostoses
Nikolaus Janocha	The role of PGM3 in Glycosylation-Dependent Signaling during Chondrogenesis
Esra Kilic	Frontometaphyseal Dysplasia with a novel FLNA gene mutation
Anika Salfelder	The Genetic Basis of Nievergelt Syndrome
Luca Sangiorgi	COL5A1 G530S Polymorphism: A Disease Modifying Factor in Ehlers Danlos Syndrome?

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Saturday, August 1, 2015

POSTER SESSION 4 – 10:30-11:00

Alistair Calder	Achondroplasia: Really Rhizomelic?
Andrea Merker	Growth Curves for Achondroplasia - Height, Weight, Body Mass Index and Head Circumference
Andrea Merker	Body Proportions in Achondroplasia
Ravi Savarirayan	Longitudinal Characterization of Growth and Morbidity in a Multinational Cohort of 74 Children with Achondroplasia
Lars Hagenäs	Achondroplasia Reference as a Background Matrix for Following Children with Extreme Short Stature
Nursel Elcioglu	The Skeletal Changes in Hurler's Syndrome after Bone-Marrow Transplantation
Ali Al Kaissi	A Venezuelan Family with Maroteaux-Lamy Syndrome and Sanfillipo Syndrome
Mustafa Kılıç	Radiogical Findings in Mucopolysaccharidosis Type VI Patients
Chong Kim	Early Enzyme Replacement Therapy in Mucopolysaccharidosis VI: Results of a Long-Term Follow-Up of Brazilian Siblings
Ali Al Kaissi	Early Senile Ankylosing Vertebral Hyperostosis in a Patient with Galactosialidosis
Nursel Elcioglu	Mucolipidosis III- Gamma Patients
Asuman Koparır	Mucolipidosis Type III Gamma: Longitudinal Observations of Four Patients Homozygous GNPTG Mutations
Mehmet Emre Atabek	A Rare Case of Mucopolysaccharidosis Required Hip Arthroplasty in a 8-Year-Old Boy with Morquio Syndrome
Alexey Baindurashvili	Comprehensive Orthopaedic Management of Patients with Skeletal Dysplasias