3as Jornadas de Doenças Ósseas Raras
Meeting on Rare Bone Diseases

Segunda-feira / Monday, 06.02.2022
10:30-11:000 – **Comunicações orais – Casuísticas e Casos Clínicos I**
Oral communications – Clinical Cases I
Moderation: *Renata Oliveira, Pedro Figueiredo*

◊ *C1. Tibial dysplasia – Etiology does matter.* (5' + 1')

Emanuel Homem, João Cabral, Inês Balacó, Cristina Alves
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◊ *C2. Pregnancy and breast feeding in 3M syndrome* (5' + 1')

Ana Grangeia[1], Inês Castro[2], Francisco Salgado-Seixa[3], Paulo Pereira[4], Cláudia Falcão-Reis[4]
[1] Medical Genetics Service, Centro Hospitalar Universitário de São João (CHUSJ), Portugal; [2]Medical Genetics Department, Centro de Genética Médica Doutor Jacinto Magalhães, Centro; Hospitalar Universitário do Porto (CHUPorto), Portugal; [3] Anesthesiology Department, Centro Hospitalar Universitário do Porto (CHUPorto), Portugal; [4] Maternal-Fetal Medicine Department, Centro Materno-Infantil do Norte, Centro Hospitalar; Universitário do Porto (CHUPorto), Portugal.

◊ *C3. TBCE-associated bone dysplasia? The importance of clinical suspicion in skeletal dysplasias.* (5' + 1')

Maria Abreu[1], Márcio Cardoso[2], Cláudia Falcão Reis[1]
[1]Medical Genetics Department, Centro de Genética Médica Jacinto de Magalhães (CGMJM), Centro Hospitalar Universitário do Porto (CHUPorto); [2] Neurology Department, Hospital de Santo António, Centro Hospitalar Universitário do Porto (CHUPorto)

◊ C4. Pfeiffer Syndrome: a family report illustrating a highly variable expressivity related to FGFR1 p.Pro252Arg variant. (5' + 1')

Mafalda Melo[1],, Susana Lemos Ferreira[1],*, Maria João Laje[2], Ema Leal[2], Daniel Virella[2], Margarida Venâncio[1], Rui Gonçalves[1]*
[1]Serviço de Genética Médica, Área de Pediatria, Hospital Dona Estefânia, Centro Hospitalar Universitário de Lisboa Central, Lisboa, Portugal; [2] Unidade de Cuidados Intensivos Neonatais, Área de Pediatria, Hospital Dona Estefânia, Centro Hospitalar Universitário de Lisboa Central, Lisboa, Portugal

◊ *C5. Osteochondromas: Multiple Problems.* (5' + 1')

Ana Rita Cavaca, Carla Carreço, Inês Balacó, Sérgio B. Sousa, Cristina Alves
Serviço de Ortopedia Pedátrica do Hospital Pediátrico – CHUC, EPE – Coimbra

Terça-feira / Tuesday, 07.02.2022
10:30-11:000 – Comunicações orais – Casuísticas e Casos Clínicos II
Oral communications – Clinical Cases II (OI and bone fragility)
Moderation: *Karen Heath; Patricia Dias*

◊ *C6. COL1A2 multiexon deletion in Osteogenesis Imperfecta type 2 – clinical case* (5' + 1')

Daniela Oliveira[1,2,3], Pedro Almeida[1], Rita Cequeira[4], Silvia Modamio-Hoybjor[5,6], Sérgio B. Sousa[1,2,3], Karen E. Heath[5,6,7], Fabiana Ramos[1,8]
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◊ *C7. Autosomal-Recessive Mutations in MESD – A Clinical Case of Severe Osteogenesis Imperfecta* (5' + 1')

Vera MF Santos[1], Ana Beleza-Meireles[1], Lina Ramos[1], Jorge Seabra[2], Pedro Cardoso[2], Luísa Diogo[3,4], Shahida Moosa[5], Bernd Wollnik[5], Jorge M Saraiva[1,4,6], Sérgio B Sousa[1,4,7]
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◊ *C8. Caffey disease: clinical case with striking neonatal presentation* (5' + 1')

Peixoto D[1], Correia L[1], Almeida S[1]; Figueiredo S[1], Tarquini O[2], Estanqueiro P[3], Modamio-Hoybjor S[4], Heath KE[4,5], Sousa SB[6,7]
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◊ *C9. Fibrous Dysplasia: case series and influence on quality-of-life* (5' + 1')

Emanuel Homem, Inês Balacó, Marcos Carvalho, João Cabral, OlianaTarquini, Pedro Sá Cardoso, Tah Pu Ling, Cristina Alves
Serviço de Ortopedia Pediátrica do Hospital Pediátrico – CHUC, EPE.

◊ *C10. Assistive products prescription and fibrous dysplasia: a case report* (5' + 1')

Lurdes Rovisco Branquinho, Francisca Melo Ferreira, João Nuno Malta, Ana Margarida Ferreira
Centro Hospitalar e Universitário de Coimbra

Terça-feira / Tuesday, 07.02.2022
11:30-12:50 – Comunicações orais – Casuísticas e Casos Clínicos III / Oral communications Clinical Cases III
Moderation: *Claúdia Reis, Paulo Coelho*

◊ C11. Achondroplasia case series: experience of a paediatric multidisciplinary clinic (8' + 2')

Luana Silva[*1] & Flávia Belinha*[*1], Núria Madureira[2], Iolanda Veiros[3], Pedro Figueiredo[3], Joana Ribeiro[4], Gustavo B. Soares[5], Beatriz Ramada[6], João E. Moura[6], João Cabral[7], Ana B. Roseiro[8], Alice Mirante[1,10], Sérgio B. Sousa[9,10]*

[1] Paediatric Endocrinology, Diabetes and Growth Unit, Hospital Pediátrico, Centro Hospitalar e Universitário de Coimbra, Portugal (HP-CHUC); [2] Laboratory of Sleep and Ventilation, HP-CHUC; [3] Paediatric Rehabilitation Unit, HP-CHUC; [4] Center for Child Development-Neuropaediatrics Unit, HP-CHUC ; [5] Neurosurgery Unit, HP-CHUC; [6] Department of Otorhinolaryngology, CHUC; [7] Department of Paediatric Orthopaedics, HP-CHUC; [8] Institute of Orthodontics, Faculty of Medicine of the University of Coimbra, University of Coimbra; [9] Medical Genetics Unit, HP-CHUC; [10] Multidisciplinary skeletal dysplasia team coordinators, HP-CHUC, European Reference Network on Rare Bone Diseases (ERN-BOND-CHUC). *Co-first authors

◊ C12. Progressive pseudorheumatoid dysplasia: the possibility of misdiagnosis (5' + 1')

M. Tomásio Neves[1], P. Dias[1] , S. Modamio-Høybjør[2], K. E. Heath[2], A. B. Sousa[1]

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◊ C13. Therapeutic Approach of Infantile Scoliosis in Osteopathia Striata with Cranial Sclerosis: revisiting a Clinical Case (5' + 1')

Maria Pia Monjardino, Pedro Cardoso, João Cabral, Joaquim Sá, Sérgio B. Sousa, Oliana Tarquini, Tah Pu Ling

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◊ C14. Intrafamilial and intragenotypic variability of skeletal ciliopathies: a case report of the Elis-van Creveld syndrome. (5' + 1')

Jorge Diogo Da Silva[1,2,3], Nataliya Tkachenko[1], Ana Maria Fortuna[1,4], Ana Rita Soares[1]

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◊ C15. Trevor disease – conservative vs surgical treatment. (5' + 1')

Sónia Rodrigues, João Cabral, Inês Balacó, Cristina Alves

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◊ C16. Recessive Multiple Epiphyseal Dysplasia – Clubfoot as a Trigger. (5' + 1')

João Pedro Marques, Marcos Carvalho, Inês Balacó, Janet Pereira, Sérgio Sousa, Cristina Alves

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◊ C17. Recessive multiple epiphyseal dysplasia case series: same SLC26A2 variant, different clinical features. (5' + 1')

Marta Marques[1], Daniela Oliveira[1,2,3], Sofia Maia[1,2,3], João Cabral[4,7], Inês Balacó[4,7], Cristina Alves[4,7], Janet Pereira[5], Karen E. Heath[6,7,8], Silvia Modamio-Hoybjor[6,7], Alice Mirante[9], Sérgio B. Sousa[1,2,3,7], Jorge M. Saraiva[1,3,10]

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◊ C18. CANT1-related skeletal dysplasia: case report of two unrelated Portuguese patients with Desbuquois dysplasia, Kim variant / MED7.. (5' + 1')

Ariana C. Mendes[1], Alice Mirante[2], João Cabral[3], Núria Madureira[4], Pedro Figueiredo[5], João E. Moura[6], Sofia Maia[1,7,8], Silvia Modamio-Hoybjor[9,10], Jorge M. Saraiva[1,8,12], Karen E. Heath[9,10,11], Sérgio B. Sousa[1,7,8]

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◊ C19. Ectopia cordis: historical case report of a rare congenital heart and sternal defect found in the Coimbra University Hospitals archives. (5' + 1')

Vítor M.J. Matos[1], Vasco Góis[2], Carlos Branco[2], Carina Marques[1,3]

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◊ C20. Management of multiple orthopaedic problems in a patient with recessive Larsen Syndrome. (5' + 1')

Andreia Moreira, Inês Balacó, Marcos Carvalho, Pedro Sá Cardoso, Tah Pu Ling, Sérgio B. Sousa, Cristina Alves

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◊ C21. Bilateral Knee Osteochondritis Dissecans within an Aggrecanopathy Case. (5' + 1')

Carla Carreço, Maria Alice Mirante, Karen E. Heath, Sérgio B. Sousa, João Cabral

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