1as Jornadas de Doenças Ósseas Raras

MEETING ON RARE BONE DISORDERS

COIMBRA, 16 e 17 de fevereiro 2018

Sexta-Feira / Friday, 16.02.2018
16:15-18:00 ◊ **Casuísticas e Casos Clínicos – I / Clinical cases – I**
Moderadores: Patrícia Dias, Iolanda Veiros

◊ C1. Novel homozygous *GPX4* mutation in a child with Sedaghatian-type spondylometaphyseal dysplasia (5+1 min)

Oana Moldovan*, Mariana Soeiro e Sá*, Patrícia Dias, Ana Berta Sousa.

*Serviço de Genética Médica, Departamento de Pediatria, Hospital de Santa Maria, Centro Hospitalar Lisboa Norte, Centro Académico de Medicina de Lisboa, Lisboa, Portugal. *Ambos os autores contribuíram igualmente para este trabalho*

◊ C2. National study on cleidocranial dysplasia – clinical and molecular characterization of 15 Portuguese patients (10+2 min)

Sofia Fernandes[1], Catarina Machado[2], Ana Rita Soares[3], Miriam Aza-Carmona[4], Renata Oliveira[1,5], Carla Pinto Moura[5], Pedro Louro[1,6], Lina Ramos[1,6], Ana Fortuna[3,7], Juliette Dupont[2], Jorge M Saraiva[1,8], Karen E. Heath[4], Sérgio B, Sousa [1,9]

[1] Medical Genetics Unit, Hospital Pediátrico, Centro Hospitalar e Universitário de Coimbra (CHUC); [2] Serviço de Genética Médica, Departamento de Pediatria, Centro Hospitalar Lisboa Norte, Hospital de Santa Maria, Centro Académico de Medicina de Lisboa; [3] Serviço de Genética Médica Dr. Jacinto Magalhães, Centro Hospitalar do Porto; [4] I Institute of Medical and Molecular Genetics (NGEMM), IdiPAZ and Skeletal dysplasia multidisciplinary Unit (UMDE), Hospital Universitário La Paz, Madrid, Spain and CIBERER, ISCIII, Madrid; [5] Serviço de Genética Médica, Centro Hospitalar de São João, Porto; [6] Faculty of Health Sciences, Universidade da Beira Interior, Covilhã; [7] UMIB - Unidade Multidisciplinar de Informação Biomédica, Portugal; [8] University Clinic of Pediatrics, Faculty of Medicine, Universidade de Coimbra; [9] University Clinic of Genetics, Faculty of Medicine, Universidade de Coimbra, Portugal.

◊ C3. Osteogenesis imperfecta type VI: a report of two cases due to a novel mutation in *SERPINF1* gene (8+2 min)

André M. Travessa, Patrícia Dias, Fátima Godinho, Manuel Cassiano Neves, Karen E. Heath, Ana Berta Sousa

Serviço de Genética Médica, Departamento de Pediatria, Hospital de Santa Maria, Centro Hospitalar Lisboa Norte, Centro Académico de Medicina de Lisboa, Lisboa, Portugal.

◊ C4. Osteogenesis imperfecta type V – report of 4 Portuguese patients (8+2 min)

João Alves, Juliette Dupont, André Travessa, Patrícia Dias, Heloísa Santos, Ana Berta Sousa

Serviço de Genética Médica, Departamento de Pediatria, Hospital de Santa Maria, Centro Hospitalar Lisboa Norte, Centro Académico de Medicina de Lisboa, Lisboa, Portugal.

◊ C5. Familial case of mild spondyloepiphyseal dysplasia congénita (5+1 min)

André M. Travessa, Patrícia Dias, Ana Berta Sousa

Serviço de Genética Médica, Departamento de Pediatria, Hospital de Santa Maria, Centro Hospitalar Lisboa Norte, Centro Académico de Medicina de Lisboa, Lisboa, Portugal.

◊ C6. The type II collagenopathies spectrum – 3 case reports: spondyloepiphyseal dysplasia congenita; spondyloepimetaphyseal dysplasia, Strudwick type; and mild spondyloepiphyseal dysplasia with early-onset osteoarthritis (10+2 min)

Sérgio B. Sousa[1,2,] Karen E. Heath[3], Joana Rosmaninho-Salgado[1], Janet Pereira[4], Jorge M. Saraiva[1,5]

[1] Medical Genetics Unit, Hospital Pediátrico, CHUC; [2] University Clinic of Genetics, Faculty of Medicine, Universidade de Coimbra; [3] INGEMM, UMDE, Hospital Universitário La Paz, Madrid, Spain and CIBERER, ISCIII, Madrid; [4] Unidade de Hematologia Molecular, Serviço de Hematologia, CHUC; [5] University Clinic of Pediatrics, Faculty of Medicine, Universidade de Coimbra

◊ C7. Bilateral osteochondritis of the interphalangeal joint of the hallux (5+1 min)

Luís Machado, Ana Daniela Pereira, Vítor Rodrigues, Carlos Ferreira, António Sá, Sérgio Figueiredo

Serviço de Ortopedia 2, Centro Hospitalar de Leiria

◊ C8. Opsismodysplasia – report on long-term follow-up of a previously described and two new Portuguese cases (10+2 min)

Pedro Almeida[1,2], Heloísa Santos[3], Jorge M. Saraiva[1,4], Jorge Seabra[5], Cláudia Reis[1], Margarida Venâncio[1,6], Karen E. Heath7, Valérie Cormier-Daire[8], Sérgio B. Sousa[1,6]

[1] Medical Genetics Unit, Hospital Pediátrico, CHUC; [2] Faculty of Health Sciences, University of Beira Interior, Covilhã; [3] Serviço de Genética Médica, Hospital de Santa Maria, Lisboa; [4] University Clinic of Pediatrics, Faculty of Medicine, University of Coimbra; [5] Serviço de Ortopedia Pediátrica, Hospital Pediátrico, CHUC; [6] University Clinic of Genetics, Faculty of Medicine, University of Coimbra; [7] INGEMM, UMDE, Hospital Universitário La Paz, Madrid, Spain and CIBERER, ISCIII, Madrid; [8] Department of Medical Genetics, INSERM U1163, Université Paris-Descartes, Institut Imagine, Hôpital Necker-Enfants Malades, Paris, France.

◊ C9. Familial case of spondyloepiphyseal dysplasia, Kimberley type, with a novel splicing mutation in Aggrecan gene (5+1 min)

Mário N. Laço[1,2], Isabel Dinis[3], Jorge M Saraiva[1,4], Lucía Sentchordi-Montané[5], Karen Heath5, Sérgio B Sousa[1,6]

[1] Medical Genetics Unit, Hospital Pediátrico, CHUC; [2] Faculty of Health Sciences, University of Beira Interior, Covilhã; [3] Unidade de Endocrinologia Pediátrica, Hospital Pediátrico, CHUC; [4] University Clinic of Pediatrics, Faculty of Medicine, University of Coimbra; [5] INGEMM, UMDE, Hospital Universitário La Paz, Madrid, Spain and CIBERER, ISCIII, Madrid; [6] University Clinic of Genetics, Faculty of Medicine, University of Coimbra.

Sábado / Saturday, 17.02.2018
11:45-12:45 ◊ **Casuísticas e Casos Clínicos – II: Raquitismos / Clinical cases – II: Rickets**
Moderadores: Clara Gomes, José Esteves da Silva

◊ C10. X-linked hypophosphatemic rickets: experience of a Pediatric Nephrology Unit (10+2 min)

Fábia Mota[1], Nuno Oliveira[1], Joana S. Caetano[2], Carmen do Carmo[1], Carolina Cordinhã[1], Clara Gomes[1], António J. Correia[1]

[1]Unidade de Nefrologia Pediátrica, [2]Unidade de Endocrinologia Pediátrica, Hospital Pediátrico, CHUC

◊ C11. Our experience in X-linked hypophosphatemia (10+2 min)

Mariana Soeiro e Sá, André Travessa, Patrícia Dias, Oana Moldovan, Ana Medeira, Heloísa Santos, Ana Berta Sousa.

Serviço de Genética Médica, Departamento de Pediatria, Hospital de Santa Maria, Centro Hospitalar Lisboa Norte, Centro Académico de Medicina de Lisboa, Lisboa, Portugal.

◊ C12. X-linked hypophosphatemia – case report (10+2 min)

Patrícia Miranda[1], Fábia Mota[2], Joana Salgado[3], Carolina Cordinhã[4], Cármen Carmo[4], Clara Gomes[4]

[1]Serviço de Pediatria, Centro Hospitalar de Leiria; [2]Serviço de Pediatria, Centro Hospitalar de Tondela Viseu, [3]Serviço de Genética Médica, Hospital Pediátrico, CHUC; [4]Unidade de Nefrologia Pediátrica, Hospital Pediátrico, CHUC

◊ C13. Two siblings with a form of vitamin-D-dependent rickets (10+2 min)

Nuno Oliveira[1], Patrícia Miranda[1], Carolina Cordinhã[1], Carmen Carmo[1], Sérgio Sousa[2], Karen Heath[3], Cristina Alves[4], Clara Gomes[1], António Jorge Correia[1]

[1]Unidade de Nefrologia Pediátrica, Hospital Pediátrico, CHUC, [2]Serviço de Genética, Hospital Pediátrico, CHUC; [3]Institute of Medical and Molecular Genetics, INGEMM, Hospital Universitario La Paz, Madrid; [4] Serviço de Ortopedia, Hospital Pediátrico, CHUC.

Sábado / Saturday, 17.02.2018
15:50-16:30 ◊ **Casuísticas e Casos Clínicos – III / Clinical cases – III**
Moderador: Manuel Salgado

◊ C14. Hypophosphatasia: report on two Portuguese cases (8+2 min)

Joana Rosmaninho-Salgado[1], Ana Beleza[1], Gabriel Matos[1],Jorge Seabra[2], Jorge Saraiva[1,3], Sérgio B Sousa[1,4]

[1]Medical Genetics Unit, Hospital Pediátrico, CHUC; [2]Serviço de Ortopedia, Hospital Pediátrico, CHUC. [3]University Clinic of Pediatrics, Faculty of Medicine, University of Coimbra; [4]University Clinic of Genetics, Faculty of Medicine, University of Coimbra, Portugal.

◊ C15. New recessive mutation in *LTPB3* in a family with short stature and amelogenesis imperfecta (8+2 min)

Patrícia Dias, André Travessa, Ana Medeira, Oana Moldovan, Ana Berta Sousa

Serviço de Genética Médica, Departamento de Pediatria, Hospital de Santa Maria, Centro Hospitalar Lisboa Norte, Centro Académico de Medicina de Lisboa, Portugal.