

BONE DYSPLASIAS

2026

SIMPÓSIO DE
DISPLASIAS
ÓSSEAS

21 a 23 Maio

Comunicações Orais - Casuísticas e Casos Clínicos I | *Oral Communications - Case Series and Clinical Cases I*

Short stature with minor skeletal abnormalities (total duration: 22 mins)

1. NPR2-Related Short Stature from a Tertiary Center: Phenotypic Spectrum with Focus on Monoallelic Cases (6 mins) - *Silva, Leandro Augusto (Endocrinology Department, IPOCFG; Unit of Paediatric Endocrinology, Diabetes and Growth, Paediatric Hospital, ULS of Coimbra)*
2. Short Stature with Compound Heterozygous CUL7 Variants: A Possible Attenuated Phenotype of 3M Syndrome (4 mins) - *Sá, Rosália (Department of Human Genetics, University Hospital Centre São João, Local Health Unit of São João; Department of Microscopy, Laboratory of Cell Biology, School of Medicine and Biomedical Sciences (ICBAS), University of Porto; Unit for Multidisciplinary Research in Biomedicine (UMIB), School of Medicine and Biomedical Sciences (ICBAS), University of Porto; ITR - Laboratory for Integrative and Translational Research in Population Health)*
3. Fifth Family with FLNB-Related Short Stature: a New Gene for Idiopathic Short Stature with Minor Skeletal Anomalies (4 mins) - *Alves, Rodrigo (Medical Genetics Department, ULS Santa Maria)*

Discussion: 8 mins

Osteogenesis Imperfecta (total duration: 36 mins)

4. Osteogenesis Imperfecta Type III and Femoral Neck Fracture – a Case Report - (4 mins) - *Cohen, Pedro (Faculdade de Medicina, Universidade Lisboa)*
5. Osteogenesis Imperfecta Diagnosed After an Apparent Isolated Femoral Fracture in a Newborn: Could We Have Done Better? (4 mins) - *Pinho, Simão (Department of Pediatric Orthopaedics, Hospital Pediátrico de Coimbra, ULS of Coimbra)*
6. Hastes Telescópicas Fassier-Duval no Tratamento da Osteogénese Imperfeita: Experiência Clínica em Dois Casos Pediátricos (4 mins) - *Santos, João (ULSB; ULSAC)*
7. COL1A1-Related Ehlers-Danlos Syndrome: Expanding Phenotypic Spectrum with Overlap Features of Osteogenesis Imperfecta (4 mins) - *Cardoso, Diana (Serviço de Genética Médica, Centro de Genética Médica Doutor Jacinto Magalhães, Clínica de Genética e de Patologia, Unidade Local de Saúde de Santo António)*
8. O Crescimento de um jovem com osteogénese imperfeita (4 mins) - *Lopes, Graça (Serviço Ortopedia, ULSSM, Hospital Santa Maria)*

Discussion: 14 mins
