

Juntar a comunidade em prol da inovação.
Bringing the community together towards innovation.

BONE DYSPLASIAS

2026

SIMPÓSIO DE
DISPLASIAS
ÓSSEAS

21 a 23 Maio

Comunicações Orais - Casuística e Casos Clínicos III | Oral Communications - Case Series and Clinical Cases III

Spine in skeletal dysplasias (total duration: 20 mins)

1. TBX6-associated Hemivertebrae due to Compound Inheritance of a Truncating Variant and a Hypomorphic Haplotype: a Mild Non-syndromic Presentation (4 mins) - *Jesus, Sara (Serviço de Genética Médica, Centro de Genética Médica Doutor Jacinto Magalhães, Clínica de Genética e de Patologia, Unidade Local de Saúde de Santo António EPE)*
2. The Hidden Risk of Spinal Cord Stenosis in CHST3-Related Recessive Larsen Syndrome (4 mins) - *Raposo, Sara (Department of Pediatric Orthopaedics – Hospital Pediátrico de Coimbra)*
3. Natural History of Spine in Achondroplasia and its Management Strategy (6 mins) - *Barreal Vega, Carmen (Fundación Alpe Acondroplasia)*

Discussion: 6 mins

Multidisciplinary care in ultra-rare skeletal dysplasias and diseases affecting the skeleton (total duration: 20 mins)

4. SLCO2A1-Related Primary Hypertrophic Osteoarthropathy: The Role of Early Diagnosis in Enabling Effective Etoricoxib Treatment (4 mins) - *Bacalhau, Mafalda (Medical Genetics Department, Unidade de Saúde Local de Santa Maria)*
5. Opsismodysplasia – From Genetics to Patient Care! (4 mins) - *Rodrigues, Joana Correia (Department of Pediatric Orthopaedics, Hospital Pediátrico de Coimbra, ULS Coimbra)*
6. Hypohidrotic Ectodermal Dysplasia in Pediatric Age: Multidisciplinary Challenges with Emphasis on Early Orofacial Rehabilitation (4 mins) - *Martins Coelho, Marta (Universidade de Lisboa, Faculdade de Medicina Dentária, UICOB)*

Discussion: 8 mins

Skeletal dysplasias with major extremities involvement (total duration: 24 mins)

7. From Brachydactyly to Symphalangism: Exploring Genotype–Phenotype Correlations in GDF5- and NOG-Related Digital Malformations (6 mins) - *Mendes C, Ariana (Medical Genetics Department, Hospital Pediátrico de Coimbra, Unidade Local de Saúde de Coimbra)*
8. Acrodysostosis: Clinical Challenges and Implications for the Dental Treatment in Pediatric Patients (4 mins) - *Nascimento, Inês (Universidade de Lisboa, Faculdade de Medicina Dentária, Dep. Odontopediatria)*

Juntar a comunidade em prol da inovação.
Bringing the community together towards innovation.



BONE DYSPLASIAS

2026

SIMPÓSIO DE
DISPLASIAS
ÓSSEAS

21 a 23 Maio

9. Acromesomelic Dysplasia, Maroteaux Type: Clinical and Molecular Characterisation of Three Cases (6 mins) - Santos, Mafalda Saraiva (Medical Genetics Department, Bone dysplasia multidisciplinary team (ERN BOND), Hospital Pediátrico de Coimbra, Unidade Local de Saúde de Coimbra; Clinical Academic Center of Coimbra, Hospital Pediátrico de Coimbra, Unidade Local de Saúde de Coimbra)

Discussion: 8 mins
